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**Syndrome without a name? The experience of living without a diagnosis for parents of  
disabled children**

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## Abstract

This thesis explores the experiences of parents with disabled children living without a diagnosis. Through thematic and narrative analysis of an in-depth qualitative interview study with 26 parents of disabled children, and by considering absent diagnosis in the context of sociological and other relevant theory, this thesis contributes to knowledge about diagnosis and about the experiences of families of disabled children without a diagnosis. The process of diagnosis, categories of diagnosis, and the consequences of living without a diagnosis are examined. Using interview data, including parent narratives and personal reflections, this thesis tells multiple stories revealing a play of diagnosis journeys: that of the parent participants living without a diagnosis; that of the researcher's exploration of diagnosis; and that of the sociological significance of diagnosis. The hermeneutic journey of the literature review process is described, as the domains of sociology and medicine have shifted shape over the years of the study. I make sense of the sociological relevance of the empirical data generated, with a particular focus on the sociology of diagnosis, ethnographic studies of paediatric genetic diagnosis, and research with families with disabled children. Despite the estimated high prevalence of disabled children without a diagnosis, there is, as yet, little research with families and to date absent diagnosis in this context has not been considered by the sociology of diagnosis. That the absence of diagnosis can hold a mirror to diagnosis, and how absent diagnosis acts to expose the *meanings* of diagnosis, is proposed. And further, to know the nucleus of diagnosis we must look at what happens in its absence, considering the space non-diagnosis leaves and the differentiability on which diagnosis abuts. I embrace a natural history approach to methodology describing the methodological journey. Further depth is added to thematic data analysis by using a narrative approach to consider parents' stories, and by punctuating the thesis with interludes of self-reflexive accounts of the researcher's own story of living without a diagnosis.

Key themes from the thematic and narrative analysis are reported: parents commonly conducted an intensive quest for diagnosis; perpetuity was a feature of this process of diagnosis, although searching for and interest in diagnosis commonly decreased over time; parents were active contributors to the process of diagnosis; parents had difficulty making sense of living without a diagnosis, and themes of *fracture* and *deferral* were identified; parents' narratives had features of the chaos, quest and restitution typologies reported by

Arthur Frank (1995), with an unresolved quest narrative as the core typology across parent accounts; a common metaphor of *stasis* of movement was identified in parent narratives. Key areas parents perceived not having a diagnosis had impacted on were: aetiology (not knowing what caused their child's disability and what the risk of recurrence was); prognosis (not knowing what to expect in the future from their child's health and development); access to support and services (including formal services and informal parent-to-parent support); and managing social interaction (how to describe their child's disability to others). Living without a diagnosis has material effects and the study's findings are relevant for theory and practice in and beyond medicine.

## **Lay Summary**

Many families with disabled children do not have a diagnosis. Some studies suggest as many as 50% of disabled children do not have a specific diagnosis, even though their difficulties may be significant. There has been very little previous research looking at how parents may experience living without a diagnosis. This thesis considers the experiences of living without a diagnosis for parents of disabled children. Interviews were carried out with 26 parents. In these interviews parents were asked to share their own story and answered questions about not having a diagnosis. Their responses were then considered in terms of the process of trying to obtain a diagnosis for their child, and the impact of not having a diagnosis. The kind of stories parents' told about not having a diagnosis was also considered. This thesis may tell us more about what a diagnosis may mean for parents and what not having a diagnosis may be like.

It was found that parents often carried out a very intensive search for diagnosis and actively contributed to the diagnosis process. Their interest in diagnosis commonly reduced over time, although their search did not end. Not having a diagnosis for their child often made it difficult for parents to make sense of their experience. Parents also reported that not having a diagnosis had an effect on how they thought about what caused their child's disability and what to expect in the future. Other key areas parents' identified as being affected by not having a diagnosis were difficulties with describing their child's disability to others and difficulties accessing support and services. This included informal parent-to-parent support and access to health, social work and education services for their child or family.

This thesis also considers concepts and areas of literature relevant to the sociological study of diagnosis, including the newly emergent sociology of diagnosis. There is also a specific focus on the process of genetic diagnosis, which is felt to be increasingly relevant in the diagnosis of childhood disability. This thesis also takes a narrative approach by gathering and analysing parents' narratives and by reflecting on the researcher's own story.

## **Declaration**

I declare:

That I have composed this thesis and that this work is my own

That the work has not been submitted for any other degree or professional qualification  
except as specified

Nicola Coates-Dutton

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I would like to express my thanks and gratitude to the parents who participated in this research, for sharing their stories with me.

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I woke to a shout:  
'I am alpha and Omega!'  
Rocks and a few trees  
Trembled in their own country  
I ran and an absence bounded beside me.

*Ted Hughes, Gog*

All I can do is tell the truth. No, that isn't so — I have missed it.  
There is no truth that, in passing through awareness, does not lie.

But one runs after it all the same.

*Jacques Lacan, The Four Fundamental Concepts of Psychoanalysis*

Always keep Ithaca in your mind.  
You are destined to arrive there.  
But don't hurry your journey at all.  
Far better if it takes many years,  
and if you are old when you anchor at the island,  
rich with all you have gained on the way,  
not expecting that Ithaca will give you wealth.  
Ithaca has given you a beautiful journey.  
Without her you would never have set out.  
She has no more left to give you.  
And if you find her poor, Ithaca has not mocked you.  
As wise as you have become, so filled with experience,  
you will have understood what these Ithacas signify.

Barry B. Powell, *Classical Myth*

For Maya, your name



## **Chapter 1      Introduction**

### **1.1      Introduction**

*Introduction*

*Research aims*

*Key terminology*

### **1.2      Research gap: research with families and theoretical literature**

### **1.3      The structure of the thesis**

### **1.4      *Starting from where you are: Personal reflections on the research topic***



## 1.1 Introduction

### *Introduction*

In this introductory chapter I will introduce the research, outlining the aims of the research and giving a brief overview of the topic, including clarifying key terminology used throughout the thesis (1.1). I identify a gap in the research relating to this topic, both in terms of research with families and theoretical literature (1.2). This lacunae in the literature was identified at the time this thesis began; and this gap remains significant. Following this, in Section 1.3, I will describe the structure of the thesis and give a brief overview of each chapter. Finally, I offer personal reflections on why the topic was chosen (1.4).

A high number of disabled children do not have a specific diagnosis. In these circumstances, there may be descriptive terminology for individual symptoms or clinical features of a child's condition, but no definitive diagnosis (Rosenthal, Biesecker and Biesecker, 2001). There is an absence of reliable and up-to-date data on the prevalence of disability in children (Blackburn, Read & Spencer, 2007), and the epidemiology of undiagnosed disability is difficult to measure due to misdiagnosis, delays in diagnosis and confusion regarding terminology. Estimates of the prevalence of children who do not have a unifying diagnosis are often projected from consultant caseloads; these estimates indicate that prevalence is high. The support organisation SWAN UK (Syndromes Without a Name) suggest that around 6,000 children are born each year with conditions that remain undiagnosed. Rosenthal, Biesecker and Biesecker (2001) report that one of the most common and unsatisfying situations encountered in medical genetics clinics is the child with multiple congenital anomalies (MCAs) for whom it is not possible to make a definitive diagnosis. It is reported that approximately 1% of new-borns have multiple congenital anomalies, with only around 40% of these receiving a diagnosis of a known syndrome (Cohen, 1997). Other authors suggest that the underlying cause of 'mental retardation' remains unknown in up to 80% of patients (Helsmoortel et al., 2014; see also Rauch et al., 2006) and that for most patients with developmental delay or intellectual disability it is difficult to identify a specific genetic (or non-genetic) cause (Miller et al., 2010).

Despite the high prevalence of undiagnosed childhood disability, this topic has received little attention from a research perspective and is currently under-theorised. This thesis explores both the lived experience of families of disabled children who do not have a diagnosis and considers absent diagnosis in the context of sociological and other theory. Through analysis of rich, qualitative data, generated through an in-depth interview study with parents of disabled children, this thesis contributes to knowledge of the experience of families of disabled children without a diagnosis. This thesis also contributes to theoretical knowledge, primarily in the field of the sociology of diagnosis and sociological and ethnographic studies of paediatric genetic diagnosis. This thesis tells multiple stories, revealing a play of diagnosis journeys: that of the parent participants living without a diagnosis; that of the researcher's exploration of diagnosis; and that of the sociological significance of diagnosis.

#### *Research aims*

The aims of the research are:

- To explore the experience of parents of disabled children who do not have a diagnosis through an in-depth qualitative interview study with mothers and fathers of disabled children.
- To understand the sociological implications of the empirical data generated by the interview study and, in conceptually reflecting on living without a diagnosis, to contribute to the sociology of diagnosis and other relevant theoretical work.

#### *Key terminology used throughout the thesis*

In this section I give a brief introduction to key terminology used throughout the thesis. I do this here so that key concepts are clarified early on in the thesis and the reader may make sense of the context in which these terms are used and the reasons for deciding to use one terminology over another.

- *Parents*            The term 'parents' is used to describe those with guardianship for their children. In all but one case, parent participants in the interview study were the biological mother or father of their child. There were no foster or adoptive parents in the study. The term 'parents' was used in the literature advertising the research to recruit participants.
- *Disabled*            Throughout the thesis I make reference to families or parents with a disabled child. The Disability Discrimination Act (DDA) 1995 definition of disability is a physical or mental impairment that has a substantial and long-term adverse effect on a person's ability to carry out normal day-to-day activities (DDA 1995 section 1.1). The literature advertising the research and introducing the research to parents used the phrase 'disabled child'. Although the DDA 1995 definition of disability was not referenced in this material, parents of children with impairment that had substantial and long-term adverse effect on their ability to carry out normal day-to-day activities volunteered to take part in the research. Parents had their own ways of describing their child's disability. In many interviews parents talked of their child's 'difficulties', therefore this phrase is also used to describe the child's disability throughout the thesis. No parent used the word 'impairment' to describe their child's disability, despite its usage in disability studies theory, therefore this word is not used throughout the thesis.
- *Diagnosis*            The phrase 'living without a diagnosis' was used in the literature advertising the research and is used throughout the thesis. Parents responded to the literature advertising the research study voluntarily, therefore self-identified as living without a diagnosis. Parents commonly were seeking a unifying diagnosis, a diagnosis that described and accounted for their child's difficulties rather than individual symptoms. They sought a specific diagnosis that was clear and meaningful. While the literature advertising the research did not specify what kind of labels were considered diagnoses or not, there was a homogeneity in the parents' perception of diagnosis as a unifying label that would account for their child's multiple difficulties.

- *Non-Diagnosis* I use the phrases 'non-diagnosis', 'absent diagnosis' and 'undiagnosed' as shortcut terminology for describing the situation of being unable to locate a specific diagnosis for a condition agreed to be medical in nature by both the family and the medical professionals involved. That is, accepted disabilities without a definitive diagnostic label. The presence or severity of a disability may not be in question; and, while there may be descriptive terminology for individual symptoms or features of the condition, there is the lack of a specific diagnosis for the overall disorder.
- *Syndrome Without a Name* I do not make reference to the phrase 'Syndrome Without a Name' when referring to disabled children without a diagnosis; rather I look at how this phrase is used by others. This phrase is primarily a lay term, having no recognition in medical classification systems, and is also used by support organisations (primarily SWAN UK) to refer to disabled children who do not have a definitive diagnosis. A discussion of the use of this phrase and of the support provided by SWAN UK can be found in Appendix 4.
- *Doctors* Parents had commonly consulted a number of different professionals, primarily health professionals, in their search for diagnosis for their child and they had different ways of referring to doctors, consultants and other professionals. Throughout the thesis, I make reference to 'doctor' as a generic term to refer to interaction parents had with doctors at different levels of training and from different specialities (for example paediatrician, neurologist, consultant). Where appropriate, for example where a parent describes in detail contact with one specific consultant, I refer to the speciality of the consultant. I also make reference to other health professionals, for example health visitors, where relevant and education professionals. Where appropriate I will specify role if it is deemed helpful for analysis.

## 1.2 The research gap: research with families and theoretical literature

A significant gap in the literature was identified at the time the research began. This gap in the literature remains, although there have been recent advances in the study of diagnosis in sociology. There are still, to date, very few studies looking at the experience of parents with disabled children who do not have a diagnosis, despite the estimated high prevalence, and absent diagnosis remains under-theorised.

Although it has been two decades since there was a distinct call for a sociology of diagnosis (Brown, 1990, 1995), only in very recent years has a systematic sociological study of diagnosis emerged (Jutel 2009, 2011a, 2011b; McGann and Hutson, 2011; see also *Social Science and Medicine* 73, 2011). Since its emergence, the sociology of diagnosis has become a dynamic yet mutable subfield of medical sociology. The key contributors invite engagement with a wide range of disciplines and discussion on the possible directions the sociology of diagnosis may take. Although there are proposals for the structure and content of a sociology of diagnosis and its future directions, the new sociology of diagnosis is still fluid, and forming shape. Within this evolving sociology of diagnosis, there is as yet no work considering the lived experience of absent diagnosis for families with disabled children, and a poverty of work considering absent diagnosis from a theoretical perspective. Although contested diagnoses and medically unexplained symptoms are explored, the situation of absent diagnosis for uncontested medical conditions is not currently considered in this literature. Furthermore, while the sociology of diagnosis is successful in theorising about diagnosis as category (in looking at the social and political forces that shape new diagnostic categories, for example) and the consequences of diagnosis for specific patient groups, it is felt that the *process* of diagnosis has received less attention. Furthermore, genetic diagnosis has rarely been described in this work, despite paediatric genetics growing in importance as a tool for explaining childhood conditions (McLaughlin and Clavering, 2011). Anspach (2011) suggests that the sociology of diagnosis demands that we examine ‘the full array of sites in which diagnosis is practiced’ (Anspach, 2011, xxiv) and consider all of the actors involved in practicing diagnosis, including parents. In considering the process of diagnosis for parents in this thesis and, in particular, the process of genetic diagnosis, it is suggested that current work in the sociology of diagnosis is utilised and extended to include additional sites in which diagnosis is practiced.

Many aspects of the experience of parenting a disabled child are well described. There is a considerable body of work looking at different aspects of caring for a disabled child and the impact of caring on parents and on parenting. The research literature, spanning the last three decades in particular, reports on the psychological, social, emotional and financial impact of caring for a disabled child or children. This wealth of literature looks at a range of issues and experiences, however there are very few studies looking specifically at the experience of parenting a child without a diagnosis. Several authors suggest the significance of diagnosis and the importance of certainty of diagnosis for parents of disabled children (Rosenthal, Biesecker and Biesecker, 2001; Graungaard and Skov, 2007; Taanila, et al., 2002; Madeo, et al., 2012; Lenhard et al., 2005; Williams, 2007; Gillman, Heyman & Swain, 2000; Gill & Maynard 1995; Brookes-Howell, 2006; Lewis, Skirton, and Jones, 2010), yet very few studies examine the impact or experience of parenting a child without a diagnosis, or look specifically at the meaning of diagnosis for parents of disabled children without a specific diagnosis.

Studies suggesting the significance of diagnosis and the importance of certainty of diagnosis for families highlight the gap in the literature in this area. Rosenthal, Biesecker and Biesecker (2001) report there are few published studies that address the experiences of families of children with an 'unidentified syndrome', and no data comparing short- or long-term outcomes for children with or without diagnoses. Other authors report that the relation between certainty of diagnosis and parental experience and coping is not well described (Graungaard and Skov, 2007) and the effects for parents of disabled children of not having a conclusive diagnosis are unclear (Lenhard et al., 2005). With reference to the use of residual diagnostic categories, Matson and Boisjoli (2007) highlight that Pervasive Development Delay Not Otherwise Specified is one of the least studied but most diagnosed of the Autism Spectrum Disorders. Similarly, Fairburn and Bohn (2005) highlight that NOS (Not Otherwise Specified) diagnoses, as residual categories, tend to be neglected by researchers. Writing recently and more specifically about families with disabled children living without a diagnosis, Lewis, Skirton and Jones (2010) suggest there are as yet very few studies detailing the psychological and social experience of parenting a child without a diagnosis. Furthermore, their review of the literature did not identify any studies conducted in the UK investigating the experience of parenting a child without a diagnosis.

A gap in the literature is identified both with reference to empirical work with families living without a diagnosis, and with reference to the theorising of absent diagnosis for conditions that are not medically contested. While recent work in the sociology of diagnosis has greatly heightened sociological focus on diagnosis, and while two studies (Rosenthal, Biesecker and Biesecker, 2001; Lewis, Skirton and Jones, 2010) specifically examine the experience of living without a diagnosis for families, the gap in literature is still felt to be significant. Given the recent diagnostic turn (McGann, 2011) in the social sciences, and the high prevalence of families with disabled children who do not have a specific diagnosis, the study of this topic is important and timely.

### **1.3 The structure of the thesis**

Here, I give an outline of the structure of the thesis and an overview of each chapter. Following this introductory chapter (chapter 1), I examine the literature in chapter 2. Chapter 2 describes the literature review as a hermeneutic journey, an iterative on-going process of understanding and interpretation. Different dimensions of diagnosis are considered (2.2), including: the historical beginnings of diagnosis, the notions of differentiation and of normalcy, categorisation and medical classification systems, and the sociology of diagnosis. Then ethnographic and sociological studies of the paediatric genetic process are reviewed (2.3), followed by a consideration of the empirical work with families with disabled children (2.4).

In the methods and methodologies chapter (chapter 3), I describe my journey to find the right methodologies for carrying out the interview study (3.2). I then look at the ethical considerations of the study (3.3), before detailing the research design methods, including how the participants were recruited and the design of the study (3.4). Methods of data analysis are then described, with reference to producing transcripts and developing themes (3.5). I finish this chapter with personal reflections on the process of research and events in my own family life at the time of the study.

I then present the findings of the research across six data analysis chapters (chapters 4 to 9). The first of these chapters looks at the process of diagnosis for parents (chapter 4), after which I consider parents' narratives of living without a diagnosis (chapter 5), before considering the perceived consequences of absent diagnosis for parents (chapters 6 – 9). In chapter 4, I examine the parents' quest for diagnosis, their process of seeking a diagnosis for their child. I look at the intensity of the quest for a diagnosis and examine how they searched for a diagnosis. I consider how parents were agentive in the search for diagnosis and how parents made sense of diagnosis. In doing so, I explore how parents sought others to help in the search for diagnosis and I look at the triggers for resuming, increasing, decreasing or stopping a search for diagnosis and how the quest for diagnosis changed over time. How the parents understood the testing process is also considered, with a particular focus on how parents made sense of genetic testing and possible attribution, and how they perceived genetics as a changing field.

In chapter 5, I look at parent narratives of living without a diagnosis. I introduce this chapter by considering the importance of narrative and the benefits of doing narrative research. I consider the kinds of stories parents told in their interviews. I utilise and extend Frank's (1995) narrative typologies, identifying 3 typologies identifiable in parent narratives: *restitution and the new normal*, *unresolved quest*, and *times of chaos*. I suggest a dominant typology of unresolved quest and identify a core metaphor of a journey metaphor of *stasis*.

Chapter 6 is the first of 4 data chapters looking broadly at the consequences of living without a diagnosis, although it is also suggested that the lived experience of living without a diagnosis may not be so easily parsed into the distinct realms of the process of diagnosis and the consequences of absent diagnosis. In this chapter I consider how parents made sense of absent diagnosis and suggest that many parents found it difficult to understand the idea of living without a diagnosis. Non-diagnosis is suggested to have low cultural resonance and I consider that the experience of non-diagnosis may represent a loss of parents' assumptive worlds (Kauffman, 2002). I also look at how parents made sense of diagnosis as deferred, and at the importance of continuing to attend the clinic for parents.

The remaining three data analysis chapters consider the impact of absent diagnosis across all temporal domains: the past (in how parents thought about what caused their child's



difficulties), the future (in assessing risk of recurrence and in the uncertainties about their child's prognosis) and the present (in accessing services, informal support, and managing social interaction). In chapter 7, I consider primarily the temporal domain of the past in examining how not having a diagnosis had an impact on how parents thought about what caused their child's difficulties. Self-blame is considered for both mothers and fathers, along with the impact of not knowing the risk of recurrence in terms of future reproductive choices. In chapter 8, I further consider the temporal domain of the future by looking at parents' concerns about their child's prognosis in the absence of diagnosis, and how they perceived uncertainty of prognosis to be related to absent diagnosis. I also report strategies used by parents to manage living with sustained uncertainty. Chapter 9 looks at access to services, access to informal support and difficulties managing social interaction for parents in the absence of diagnosis. In doing so, this chapter is more focused on the impact of absent diagnosis on the temporal domain of the present in looking at the day-to-day lives of the parents. I report that many parents felt access to services was affected by not having a diagnosis, and that there was unmet need for informal support. I suggest that the experience of living without a diagnosis may result in feelings of isolation, further compounded by the difficulties parents may have managing definitions of their child in social interaction.

The final chapter offers a discussion of the work and conclusion to the thesis. I highlight key themes identified in the research and suggest that the thesis contributes to knowledge with regards to both empirical work with families and sociological analysis of diagnosis.

#### **1.4 Starting from where you are: personal reflections on the research topic**

*I had intended to punctuate this thesis with 'interludes' telling my own story of living without a diagnosis. In the end, I underestimated this task, both for its complexity and for the ethical issues involved in revealing a personal story that is not protected by at least some degree of anonymity, making it difficult to protect the privacy of other family members. As mummy, advocate, and 'warrior' (in my husband's kind words) for my girls, I have spent years endeavouring to protect the privacy of a family that has at times seemed to carry the stamp of 'Property of the Royal Hospital for Sick Children'. My intention to tell my own story through in-depth personal reflections is referred to in the abstract of this thesis, symbolising that the process of learning over the course of a PhD can occur right up to the stage of the final draft. Instead, I have much abridged the story I intended telling. Perhaps this is akin to the decision research participants may make when they are advised in the process of informed consent that anonymity cannot be guaranteed, adjusting what they say with the knowledge that they may be recognised. In this thesis, I share short personal reflections about how I came to choose the topic for the research and the impact of being the parent of a child without a diagnosis on the progress of the research over the years. The content of these extracts were edited to remove any parts that my eldest daughter, Maya, felt uncomfortable with. As a teenager at the time of writing, she was old enough to decide what aspects of our story she was comfortable with me sharing.*

*My eldest daughter, Maya, was born in May 2000, when I was twenty-six. She was named after my mother, May; the writer, Maya Angelou; the Nepali word for love; the Sanskrit word for illusion referring to a phenomenal universe subject to differentiation and impermanence; and a pub in Kathmandu 'Pub Maya' where her dad and I first spoke to each other. I was so utterly intoxicated by my feelings for her that I didn't pay much attention when the paediatrician called her floppy. I'd never encountered the word 'floppy' as a medical term. I enjoyed two days of complete ignorance that anything was wrong, before the process began of her going 'upstairs' for some tests, a journey that was to last five months before she was able to come home, and that endured beyond this. The world had changed shape and I didn't understand its new mould. Maya was beautiful, and the mother love was joyful yet debilitating. I experienced extreme joy and chronic sorrow often within the same glance. I was a terrible pseudo-patient, demanding and anxious. During her 5 month stay in hospital, Maya had a gastrostomy placed for long-term tube feeding as her muscle tone was too low for her to swallow safely, and she also contracted whooping cough that saw her ventilated for a whole month in paediatric intensive care.*

*I'm not sure when the process of diagnosis began, I would say immediately on arriving 'upstairs' at two days old. At first, it didn't belong to me. The geneticist had given her the 'once over' and said there was 'nothing obvious'. She was tested for Prader-Willi Syndrome and a consultant suggested that often children who are floppy have 'benign congenital hypotonia' and catch up with their peers by the time they go to school. I carried this dichotomy of possibilities around for a while, the idea about it being one thing or the other. During this time I read a bit about Prader-Willi syndrome and I joined an online support group for parents of children with benign congenital hypotonia. I swung between imagining a life with locks on food cupboard doors and a child with a lifelong physical and intellectual disability, and a life with a bookworm of a child who would mature out of her development delay. One day the test for Prader-Willi came back negative and soon after that my internet searching led me to a letter to the editor of the journal *Developmental Medicine and Child Neurology* entitled 'Benign congenital hypotonia is not a diagnosis' (Thomson, 2002). And so the sands shifted again.*

*While she was still in hospital, and long after she came home, I pursued diagnosis. Something drove me to find 'the answer'. Having considered that a genetic explanation was ruled out following testing, there seemed no parameters to what may have caused her disability. Was it because I drank a beer? Took a vitamin A supplement? Touched a poisonous frog in Thailand? Everything was culprit. The triplets being delivered in the next room in the labour suite when she was born, the ones who needed my midwife's attention, they were culprit. The paediatrician who handled her a little too roughly when she was being examined after being born, he was culprit. The midwife who detangled her from her own placenta's cord around her neck, she was culprit. But most of all, I was culprit.*

*An answer was there, it was just eluding me. The line from the Ted Hughes' poem Gog came to me many times a day, 'I ran, and an absence bounded beside me'. I polled members of an 'unidentified syndrome' board online asking about their pregnancies, trying to tie threads together. Meanwhile, she came home, 144 days after being born. She came home on oxygen, with a saturation monitor and a suction machine, and a gastrostomy. At 5 months old, I was still expressing breast milk. She didn't tolerate large volumes so I would feed her 10-12 small feeds a day, expressing in between. I had no idea about her prognosis. In many ways I was just glad she was alive.*

*One day, while leaning against a radiator in the middle of winter, I considered the possibility that she wasn't going to get a diagnosis. I wondered what other parents did when faced with absent diagnosis,*

*how did other parents feel? And just like that the paradigm of my thinking shifted. I had already started a Masters in Social Research at Strathclyde University, the taught element of which I started while Maya was 5 months old and still in hospital. I was keen to learn more than I was learning from reading the short accounts of having an undiagnosed child on internet forums, so my Masters dissertation was an in-depth interview study with 6 parents of children who didn't have a diagnosis. The catalyst for gathering these stories about diagnosis was my own experience. A world I had assumed was fractured. In facing my own biographical disruption, I wondered how others experienced living without a diagnosis. In a sense, I stopped looking for a diagnosis, and started looking at diagnosis. Perhaps this thesis is my narrative repair, or that carrying out the research was a kind of coping strategy giving me an acting possibility in the absence of being able to locate diagnosis. In starting from where I was, I travelled to listen to the stories of other parents and, in doing so, this thesis began.*



## **Chapter 2      Literature Review**

### **2.1      Introduction**

### **2.2      Diagnosis**

*Differentiating*

*The beginnings of diagnosis*

*The notion of the norm*

*Categorisation and medicalisation*

*The sociology of diagnosis*

### **2.3      The process of paediatric genetic diagnosis**

*The visual culture of dysmorphology*

*Parental participation in the process of genetic diagnosis*

*Diagnosis as deferred and being kept on in the clinic*

### **2.4      Research with Families**

*Parenting a disabled child*

*Living without a diagnosis*

*Other research that considers diagnosis*

### **2.5      Conclusion**

## 2.1 Introduction

A literature review evolves over time; it is a hermeneutic journey. A deeper understanding of the research aims and questions is gained as engagement with the literature advances, and the literature review develops and expands into further relevant adjunct areas as the research is cultivated. Boell and Cecez-Kecmanovic (2010, 2014) suggest that reading, conducting research, and writing are not a linear process, but instead iterative with on-going reading important to the understanding process. This chapter reviews the literature I examined as I journeyed through a process of learning about diagnosis, a process that spanned a decade. In this time, the landscape of the literature changed.

The terrain of the literature available in 2014 is very different when compared to the beginning of the study. In 2005, at the beginning of this study, and still in 2008 when the writing up of this thesis began, there remained only a call for a sociology for diagnosis, and not yet a response. Work on the ethnography of dysmorphology was only just emerging and there were few studies looked at the process of genetic diagnosis, particularly in paediatrics. There was only one study to have looked directly at the experience of living without a diagnosis for families with 'unidentified congenital anomalies' (Rosenthal, Biesecker and Biesecker, 2001) and a scattering of research that considered the significance of diagnosis for families with disabled children. In the field of medical genetics, while the human genome had been sequenced, the ability to make novel disease discoveries in a therapeutic setting using next-generation sequencing equipment was still visionary. The landscape for categorising pathology in other domains was also shifting, as revisions for the Diagnostic and Statistical Manual of Mental Disorders DSM V were underway, although this newest version of the DSM was not published until 2013. In my own world, I was yet to discover the further significance of living without a diagnosis for my own family, as I began writing up the thesis anticipating pregnancy with my second child.

Literature during these years was gathered cumulatively, snowballing over time after an initial intensive period of exploration and gathering. Identifying the research topic was complex; different authors used different terminology to describe absent diagnosis. After identifying central authors and core journals and a number of key texts and papers, I used

reference tracking and citation analysis as a way of tracking relevant literature backwards and forwards in time. I quickly realised that the study of diagnosis involved multiple disciplines and fields, and I had soon gathered a range of theoretical and empirical work around the topic of diagnosis. However, I also quickly realised that there were significant gaps in areas that were key to the study; the call for a sociology of diagnosis by Brown in 1995 had not yet been answered, and there was no qualitative work following on from Rosenthal, Biesecker and Biesecker, (2001) study of the experience of absent diagnosis for parents of disabled children with multiple congenital anomalies. Had writing of the thesis, which began in 2008, been completed by 2009 as intended, this literature review would have been quite different. The additional time taken to complete the thesis meant additional iterations and I returned again and again to the areas of relevant literature analysing and synthesising new work. For some fields, like the sociology of diagnosis that grew exponentially within a short period of time between 2009 and 2011, this work was considerable and vital. The result of using an iterative approach was a better approximation towards more relevant literature and the ability to identify and synthesise new adjacent areas. It also, however, meant for a longer journey.

With a hermeneutic approach to the literature review, there is no final understanding of *the* relevant literature, but a continuing open-ended process in which constant re-interpretation leads to a more comprehensive understanding of relevant publications (Boell and Cecez-Kecmanovic, 2010, 2014). Systematic reviews are not necessarily suitable for humanities and social science research, as they require the research question to be set in advance (Boell and Cecez-Kecmanovic, 2010, 2014). Clear boundaries to what is relevant literature can often not be established at the beginning of social science research and understandings and perspectives gained throughout the research process may widen the circle of relevant literature. The process of analysing and synthesising literature is on-going and circuitous and, in making sense of what is relevant literature and how, we shift back and forth between the parts and the whole, creating interpretation.

This literature review is structured thematically, as I analyse relevant literature from different theoretical and research fields and, in later chapters, synthesise this knowledge with my own empirical work. Although divisions have been made between different theoretical and research fields, there is much crossover between them and other ways of



organising this review are possible. The literature relevant to research methods and to narrative research and analysis are discussed in later chapters (Chapters 3 and 6). Also, literature which is relevant only to specific data analysis results will be introduced in the appropriate data chapters. Here, I primarily introduce relevant literature about diagnosis and from the field of the newly emergent sociology of diagnosis, studies of the process of paediatric genetic diagnosis, and research with families with disabled children.

I begin this literature review by exploring literature related to diagnosis (2.2). There are multiple concepts that are useful to unpack when considering diagnosis and its absence. There are also related academic fields dedicated to the study of diagnosis, most recently the newly emergent sociology of diagnosis. As well as considering this new subfield of medical sociology and other related fields within and outside of medical sociology, this first section on diagnosis aims to examine the assumptions that are embedded in the practice of identifying and naming medical conditions and disorders of childhood. This section looks at five interconnected areas relevant to the concept and study of diagnosis: *differentiating; the beginnings of diagnosis; the notion of the norm; categorisation and medicalisation; and the sociology of diagnosis*. These areas are inextricably related to each other and studying them as distinct may conceal their reciprocal, shifting relations. I therefore consider these concepts while being self-reflexive about the risk of reifying them as distinct. In differentiating in this way between subjects relevant to the study of diagnosis, the approach in this chapter is autological. In differentiating between categories, I am splitting concepts that are different enough to find a good enough seam between them, and lumping concepts that are similar enough to make sense together.

Following on from considering these aspects of diagnosis and its study, I examine literature that looks at the process of paediatric genetic diagnosis (2.3); the majority of this work involves ethnographic study of the process of paediatric genetic diagnosis. As my research study advanced, paediatric genetics grew in significance as a tool to explain childhood illness and disability (McLaughlin and Clavering, 2011). It became increasingly pertinent to consider knowledge about new genetic technologies and advances in genetic diagnosis and to examine work looking at the process of genetic diagnosis.

Finally I consider research with families (2.4). I look at research with families with a disabled child, with families living without a diagnosis, and other research that considers diagnosis. Research with families with disabled children who do not have a specific diagnosis is commonly empirical work using qualitative methods, primarily interview studies (Gruangard and Skov, 2007; Lewis, Skirton and Jones, 2010; Makela et al., 2009; Rosenthal, Biesecker and Biesecker, 2001), although ethnographic studies of families in the genetics clinic are also reviewed here (Brookes-Howell, 2006) as well as in the section looking more specifically at paediatric genetic diagnosis. One study with parents of disabled children with and without diagnosis was a questionnaire study (Lenhard et al., 2005) and another was a mixed method (quantitative and qualitative) survey (Madeo et al., 2012). Other research exploring the importance of diagnosis for families, although not specifically relating to absent diagnosis, is also considered. The majority of this work is empirical and qualitative.

## 2.2 Diagnosis

### *Differentiating*

The word *diagnosis* is derived through Latin from the Greek *diagignōskein*, to distinguish, and *gignōskein*, to perceive (Collins English Dictionary, 2003). Perceiving things apart, differentiating, is fundamental to diagnosis. Diagnosis is relational, apart from others that it is not, and knowing about diagnosis means recognising the differential nature of how we organise our understandings about the world, which is particularly pertinent to Western culture with its biomedical drive.

As Zerubavel (1993) highlights from the beginning of his writing on making distinctions in everyday life, the biblical story of creation, like other cosmogonies, is an account of how we create order from chaos by making distinctions and thus making sense (1993, p1). Separating entities from their surroundings allows us to perceive them and, in perceiving them, make sense of them. We take a 'boundless, undifferentiated void' (Zerubavel, 1993, p1) and carve it up so that it makes sense. Where we situate the boundary lines may seem to reflect natural

categories although they divide a continuum into categories on the basis of our conventions. Boundary lines vary from one society to another and across time. We often experience boundaries as if they were parts of nature, rather than as mental constructs that allow us to make sense of our environment. Bowker and Star (2000) suggest that classifications are powerful technologies, with everyday categorising practices disappearing into the infrastructure, habit, into the realm of the taken-for-granted. The act of assigning things or people, or their behaviour and action, to categories is held to be a 'ubiquitous part' of the modern world and categories come to be naturalised (Bowker and Star, 2000, p323). Classification systems and standards 'literally saturate our environment' (2000, p38) and yet in a way that, given their naturalisation, they may become taken for granted unnoticed aspects of our lives: 'the ubiquity of classifications and standards is curiously difficult to see' (2000, p38).

The way we 'cut up the world', categorise, draw boundary lines, distinguish one thing from another, influences our social order and the way we organise our everyday life (Zerubavel, 1993, p1). We transform the natural world into a social one, by 'carving out of it mental chunks we treat as if they were discrete' (1993, p5). Yet while treating things as if they were discrete, at the same time we ignore their uniqueness and treat them as members of a particular class of objects, acts or events.

Indeed, things become meaningful only when placed in some category (Zerubavel, 1993, p5)

Thus we simultaneously differentiate and unify. We partition space and time, distinguish figure from ground (1993, p5). We have borders that are often unseen other than when visually represented on a map, and we perceive breaks in time that mark distinctions between last week and this one, yesterday and today, past from present from future. Our being involves a play of differentiating and unifying, what Zerubavel calls lumping and splitting (1993, p21).

Even our average 'everydayness' (Lefebvre, 1947) involves differentiating innumerable different things from others so that we function with relative ease. 'We are constantly negotiating life's lumps and splits' says David Weinberger (2007) writing about order and the miscellaneous in our modern digital worlds. Most of this differentiating is not done with conscious awareness but has been naturalised, and it is only by examining the distinctions

we make do we bring them to conscious awareness. Most distinctions in our average everydayness are simultaneously so naturalised that they go unnoticed, yet so significant that to ignore, refute or be unable to abide by them would significantly alter our worlds. Garfinkel (1967), with the intention of demonstrating ethnomethodology, used a teaching practice of conducting breaching experiments with his students. Such violations of social norms of behaviour highlight the ways people construct social reality (Ritzer, 2011). According to Garfinkel, these experiments are meaningful as they allow us to see the 'seen but unnoticed' background features of everyday life (Garfinkel, 1967, p118). Garfinkel's experiments concerned behaviour, breaching norms of behaviour exposed the socially standardised expectations of everyday scenes, but the concept of breaching norms in other contexts has similar effect. Here, in considering Zerubavel's work, I acknowledge that we function, perceive and understand by distinctions; they are how we think, often without thinking. These distinctions go unnoticed but we can bring them to the fore and acknowledge their nature as cultural constructs.

It is possible to identify ways of being that do not rely on differentiating. The inherent oneness of the world and universe is described in disciplines as diverse as religion and science. In Hinduism, for example, according to the *Advaita-Vedanta* (meaning non-dualism) view in Indian philosophy, man and God are the same thing. We only perceive separate selves because of *maya* (that is, illusion) (Potter, 1981; Lochtefeld, 2002). This non-dual way of being represents enlightenment. In quantum physics, a sense of oneness is also revealed. Schrodinger (1958) describes a basic oneness of the universe.

The world is given to me only once, not one existing and one perceived. Subject and object are only one. The barrier between them cannot be said to have broken down as a result of recent experience in the physical sciences, for this barrier does not exist (1958, p127)

Yet in Western culture, for the most part, we do not think and carry out our daily lives through these domains. In being social, and in thinking through a culture that privileges biomedical thought, we think through the duality of ego and believe the distinctions we make to be natural.

Diagnosis is not about oneness. With diagnosis the world is given to us twice, the one existing and the one perceived. Diagnosis is a social act and it is relational; marking boundaries between things, differentiating. Diagnosis and our fascination with diagnosis belong to a classificatory style of medicine, which is the mainstay of Western medicine. We have ego, and are distinction creators who perceive our distinctions to be true. As beings-for-themselves, whose consciousness renders us entirely different from other things (Sartre, 1958), we live relationally. Diagnosis is boundary work. As Zerebual suggests, we divide what would otherwise be continuous concepts into discreet entities, like seas and lands, and in the drive to classify we have 'transformed oceans into mental archipelagos' (Zerubavel, 1996, p62).

This is an important place to start when considering diagnosis, that it depends on a system of thought that privileges division as a way of understanding the world. Authors writing from within the newly emergent sociology of diagnosis do consider that studying diagnosis reveals fundamental aspects of the way we live in and relate to the world around us; 'Diagnosis tells us much about what we value and believe in as people; it behooves us to understand it well' (Jutel, 2011a, p139). The principal authors from the discipline also acknowledge that their focus is on modern Western medicine and highlight gaps in the sociology of diagnosis in the consideration of different systems of thought. By discussing diagnosis we are dealing with the biomedical model of the body that dominates Western medicine. Modern Western biomedicine relies on specificity, standardisation and generalizability (Rosenberg, 2002). These aspects should not be taken for granted when considering diagnosis but seen as one way of organising how we think about disease, illness, and disability. In considering diagnosis, I focus on modern Western medicine akin to other authors within the sociology of diagnosis. I acknowledge, however, that there are other ways of being and forms of medicine that may have a different dialogue with diagnosis. The parent participants in my study experience absent diagnosis in the context of modern Western medicine and the quest for a diagnosis reflects a commitment to a system of medicine that is nosologically organized, precise, certain and ever progressing. That is, a commitment to biomedicine.

Diagnosis is both a verb and a noun; it is the work carried out to solve the puzzle, and the answer to the puzzle itself. It is both process and category; the pre-existing set of categories agreed upon to designate a specific condition that is considered pathological, and also the process by which the label is applied (Blaxter, 1978). Diagnosis is at the heart of the science and art of modern medicine, the nucleus of the cell of medicine. Although a detailed history of medicine is not possible within the scope of this thesis, it is useful to identify junctures in the history of medicine in which diagnosis became organized around nosological principles; for it is these nosological principles that have carried forth and may inform how we perceive diagnosis as capable of precision and accuracy.

The ideal classification system is one that encompasses everything; classification systems are designed based on this ideal (Bowker and Star, 2000). Without classification systems, diagnosis comes undone. Therefore, a brief consideration of how medical classification systems evolved is worthwhile. It is also particularly important to acknowledge the dual meanings of diagnosis as category and process; in a study of parents living without a diagnosis, the *process* of seeking diagnosis is prolonged, and parents may have an enduring dialogue with *categories* of diagnosis as they journey through this process. Furthermore, recognising the importance of diagnosis in medicine and beyond may tell us something about the significance of its absence.

Diagnosis is crucial to the biomedical model of disease and the scientific paradigm of definitive etiology. Diagnosis organises illness (Balint, 1964) and is intrinsic to the theory and practice of medicine (Brown 1995, p38); 'the fulcrum of the medical narrative' (Jutel, 2009, p288). Diagnosis does a considerable amount of cultural work in health and medicine (Bowker and Star, 2000); leading backwards to the aetiology of the disease and forwards to treatment interventions (Arksey and Sloper, 1999). Diagnoses suggest how cultures define normality and abnormality, helping to define what is deviant and anomalous (Armstrong, 1995). Diagnoses are the suture points that connect lay experience and professionally sanctioned knowledge; 'Diagnosis matters deeply – both for therapeutic and symbolic purposes' (Fuller, 2011, p239).

Diagnosis as distinct category is a relatively recent phenomena (Anspach, 2011). Before the emergence of modern diagnosis in the mid-nineteenth century (Rosenberg, 2002), diagnoses were fluid, varying and focused on the individual patient (Anspach, 2011). The patient's subjective narrative was the focus of the medical encounter. Beginning in the 19<sup>th</sup> century the practice of diagnosis moved away from the patient's subjective narrative towards clinical signs of disease. An interpretive wall (Anspach, 2011) was created that was further augmented by the introduction of diagnostic technologies, for example the stethoscope. Interpretation of illness thus became more reliant on the clinicians technologically mediated pictures of disease than the ill person's narrative (Anspach, 2011).

Jutel (2011a) describes how diagnostic medicine learned much from biology in the early 18<sup>th</sup> century, reporting that the naturalists from Europe sailed the world at the start of the 18<sup>th</sup> century; exploring, collecting and categorising the living world. Soon, a classification movement was in full swing and taxonomy would come to be a dominant paradigm for understanding the world (Jutel, 2011).

Medicine followed the natural sciences and engaged in the great classificatory project that would underpin diagnosis (Jutel, 2011, p6)

This shift towards a classification scheme marked a turning point in the history of medicine, one from a focus on individual diverse idiosyncratic symptoms to a system that perceived diseases as comprising of symptom clusters. This paradigm shift underpins the concept of diagnosis.

Foucault's work traces the history of the shifting paradigms, or *epistemes*, of medical knowledge through the 16<sup>th</sup> to 19<sup>th</sup> centuries. Foucault, in *The Order of Things* (1970, 2001), describes a move from the age of theatre to the age of the catalogue; asserting that, through the drive to catalogue in the natural sciences, 'seeing' was privileged with sight being awarded an 'almost exclusive privilege' that was augmented by inventions like the microscope (2001, p144). Other senses become less relevant to medical knowledge. In *The Order of Things* (1970, 2001) and *The Birth of The Clinic* (1973, 2012) the late 18<sup>th</sup> century is held to be the dawning of a new empirical system. In Foucault's view, this represented a momentous shift in the structure of knowledge; 'an essential rupture in the Western world'

(2001, p55). The fascination with the botanical world was not new; the shift occurred in the *way* of seeing, not what was seen. A paradigm (the age of the theatre) of seeing that world as *show*, became a paradigm (the age of the catalogue) of seeing and organising that world as *table*. An episteme of similitude in the 16<sup>th</sup> century became an episteme of difference in the 17<sup>th</sup>-18<sup>th</sup> century Classical period; 'what has become important is no longer resemblances but identities and differences' (2001, p55). The new crucial instrumentation is representation rather than similitude, and it is in this new space of taxonomy that diagnosis flourishes.

In *The Birth of the Clinic* (1973, 2012), Foucault describes changes in the dominant approach to disease in the late 18<sup>th</sup> century as marking the birth of modern medicine. With the birth of a new type of clinic in the late 18<sup>th</sup> century so too was born a new clinical gaze (*le regard* of medicine) characterised by an increased emphasis on physical examination and observation of localized bodily disorders, where the approach had previously been more holistic and more based on the patient's own narrative. In time this new clinical gaze gave rise to a belief in the body as a stable invariant reality and so a new way of looking at the body was born, a new 'anatomical atlas' emerged (Foucault 1973, 2012). Medicine began to be conceptualized around normality of function; 'regulated more in accordance with normality than with health' (Foucault 1973, 2012, p40). The subsequent growth of diagnostic technology augmented diagnosis as an expert system of knowledge. Diagnosis of disorder, a cluster of symptoms, thus only became central to medical practice consequent to this paradigm shift in which knowledge about bodies and their pathologies changed entirely.

Foucault illustrates how the practices of medicine, particularly with reference to the practices of diagnosis, are intricately linked with wider social phenomena. He identifies that a fundamental shift in medical perception at end of 18<sup>th</sup> century brings about a revolution in the understanding of disease and illness. Disease came to be seen as pathological lesion localised to a distinct point within the body and medicine became about the search for this lesion. This new medicine relied on diagnosis and was accompanied by novel classification. In time, diseases came to be regarded as universal, natural objects existing prior to and independent of their identification and diagnosis by doctors (Morgan, Calnan and Manning, 1985). The biomedical model of disease continues to be the dominant paradigm through which we construe illness, disease and bodily disorder. Armstrong (1995) describes hospital medicine (also known as the clinic, pathological medicine, western medicine and



biomedicine) as an extensive revolution in medical thinking that has endured and extended itself over the last two centuries to become the prevailing model of medicine in the modern world (Armstrong, 1995). It is a paradigm in which diagnosis is imperative.

### *Normalcy*

The categorisation of childhood disability hinges on the notion of the norm. To understand the construction of disability and the disabled body, we must return to the construction of normalcy and the normal body (Davis, 1997). The notion of the norm is less a condition of human nature and more a characteristic of a certain kind of society, a product of a particular historical moment (Davis, 1997). Indeed the word normal only entered the English language in around 1840 (Davis, 1997). The advent of the use of statistics to read a population, in the 1830s and 1840s in Britain, was vital to the construction of the notion of the norm and the idea of the man in the middle, the norm, as imperious. 'The man in the middle' soon came to be an archetype of the middle way of life, the new ideal (Davis, 1997, p5). With the notion of the norm and the normal curve (applied to both morals and bodies) came the concept of deviation from that norm. The tool of the normal curve and the concept of normal distribution underpins the diagnosing of disease and disability. Foucault identifies the normative assumptions of early 19<sup>th</sup> century medicine, suggesting that medicine becomes regulated by the concept of normality (Foucault, 1973). In *The Birth of the clinic* (1973), and later in *Discipline and Punishment* (1977), Foucault maps the process of how bodies came to be regarded as 'normal' or not through medicine's categorising of bodies held to be outside of the norm. Categories of diagnosis are implicated in establishing and reinforcing cultural conceptualisations of normal bodies.

The norm is an entirely relative notion as Canguilhem (1989) clarifies; 'norms are relative to each in a system' (1989, p249), they have correlativity only within their own social system. Normative order characterises modern societies, with the norm being a way for a group to provide itself with a common denominator that will never refer to anything external to the group, i.e. beyond itself. The norm can never be absolute nor universal and is always time bound. McLaughlin and Coleman-Fountain (2014) draw on Canguilhem (1989) and Lester

and Paulus (2012) in suggesting that disability can be thought of as a core category in establishing the normality of persons. Rather than normality being seen to establish disability, they suggest that the normal is established by identifying what is deemed to be 'strange and ill-fitting', that is, abnormal (McLaughlin and Coleman-Fountain, 2014, p77). They also highlight that those who sit within the category of the abnormal or pathological are always in relationship to those in the category of normal. As Ewald (1990) suggests, there can be no such thing as a norm that exists in isolation for a norm never refers to anything else other than other norms on which it depends.

According to Foucault (1973), the political significance of the norm arose in concert with modes of governance that relied on population statistics and associated human sciences. Populations were monitored with a view to individuals monitoring (governing) themselves. These practices gave rise to the importance of normal development, normal behaviour, normal functioning, normal health (Nettleton, Neale and Pickering, 2012). We are assessed from birth and throughout life, with the development of the child particularly subject to surveillance (Armstrong, 1995). One of the earliest expressions of the surveillance medicine that began to emerge at the beginning of the 20<sup>th</sup> century was the problematisation of the normal, with whole populations targeted for surveillance. In particular, the medical gaze (Foucault, 1973) turned quickly on the child, with the growing child's mind and body closely observed and assessed (Armstrong, 1995). In measurement scales like height and growth charts the child is abnormal only relative to other children, and then only by degrees (Armstrong, 1995). The regular routine surveillance and measurement of the development of the child has grown over time. A child is subject to surveillance from before she is born, and as she grows, by the monitoring and measurement of her body, growth and development against norms of the population.

Davis suggests that medicine has produced the 'concept of the disabled body' (1995, p30 cited by McLaughlin and Coleman-Fountain, 2014). The concept of disability is inextricably linked to the concept of the norm. Writers from within disability studies (for example Barnes and Mercer, 2010) have drawn our attention to medicine's power to identify certain body types as lying outside measures of normality and to categorise such bodies as 'disabled'. The notion of the norm underpins the delineating of a child's development as disordered. Relative to other children in the same population, a child is found to have a disorder of

development; they are the outliers of the normal curve. The parent participants in my study, other than those identified as having an atypical experience, had children who were considered to have a pathological delay in development. The delineating of the child as having abnormal development relied on the notion of the norm. Here, I suggest that this notion of the norm underpins diagnosis and highlight authors who have considered the implications of the hegemony of normalcy and have suggested that, while 'the norm' is a concept that permeates our contemporary life, it arose at a particular historical moment and in a particular cultural context. For families of disabled children, following the identification of an abnormality in child's development a more specific diagnosis is sought. The process of diagnosis begins with delineating of the normal from the abnormal. This process then becomes a search for a more specific diagnosis, which relies on engagement with systems of medical classification. I will now consider aspects of categorisation and medical classification to add a further dimension to our understanding of diagnosis and its counterpart, absent diagnosis.

#### *Categorisation and medical classification*

Parents of children without a diagnosis experience a lengthy process of diagnosis in which they have an enduring engagement with categories of diagnosis. The search for diagnosis involves a process of considering multiple diagnoses within classification systems in order to arrive at the right diagnosis. Parents who seek a definitive diagnosis for their child are seeking a precise medical category. Although parents may not be familiar with how classification systems are structured, they will be aware of a plethora of categories that define illness and disability. Having noted the significance of the notions of normalcy and the historical beginnings of diagnosis earlier in this review of the literature, here I consider classification and categorisation and introduce literature that explores the relationship between diagnosis and classification and the structure and cultural work of classification systems, and considers medicalisation and disputed or contested diagnoses.

Jutel (2011a) suggests that diagnosis is one of medicine's most powerful classification tools. Classifying recognises difference as well as similarity, and by classifying we are putting

items together that have more in common with one another than they do with things that belong in another category. There must be enough resemblance among members of a category.

Classification is a process that relies on the abilities to distinguish same from different: clustering those things that resemble one another into a group, distancing those items that don't belong (Jutel, 2011a, p39)

Classification is seen to 'shape(s) medicine and guide(s) its practice' (Jutel, 2011b, p189). Latimer (2013) states that what makes medicine a science is 'the same as what keeps it modern; the construction of systematic forms of classification that can be used to categorize people' (Latimer, 2013, p7). Diagnosis does not work without classification. Bowker and Star (2000) examine how classification systems scaffold information structures; in doing so they look specifically at the International Classification of Diseases (ICD), an 'information infrastructure' (2000, p107) and 'international tool for standard diagnostic classification' (2000, p72), to illustrate their work. The ICD is a classification system seen to do 'work', both bureaucratic work and knowledge production (2000, p10).

Bowker and Star (2000) address the question of what happens to the cases that don't 'fit' in medical classification systems. They explore the use of 'not otherwise specified' categories that are distributed throughout the entire ICD as a means of dealing with uncertainty and ambiguity.

Since uncertainty is inevitable, and its scope and scale essentially unknowable, at least its impact will not hit a single disease or location disproportionately. Its effects will remain as local as possible; the quest for certainty is not lost, but postponed, diluted and abridged (Bowker and Star, 2000, p25)

The not otherwise specified category is exposed as a 'protean modifier' (2000, p100) throughout the classification, and one that constantly and systematically attempts to defer uncertainty and shrink ambiguity. Such 'garbage categories', as described by Bowker and Star (2000, p149), include 'an array of categories where things get put that you do not know what to do with – the ubiquitous 'other' (2000, p149). The management of residual categories is a continuous theme throughout the history of the ICD, a significant 'fault line' of this system that endeavours to formally decipher the human body.

A fracture that is constantly being redefined and changing in its nature as the plate of lived experience is subducted under the crust of scientific knowledge....The crack comes when the messy flow of bodily and natural experience must be ordered against a formal, neat set of categories (Bowker and Star, 2000, p67 – 68)

Another important focus of Bowker and Star's work is that classification systems are held to be culturally constructed; 'standards and classifications, however dry and formal on the surfaces, are suffused with traces of political and social work' (2000, p49). Bowker and Star focus on the political and social influences on the evolution of the ICD and describe the system as an 'ineluctably arbitrary way of cutting up the world' (2000, p101). In this way, their work aligns with Zerubavel's work on the creation of boundaries from the flux of human existence, boundaries that construct our social reality (Zerubavel, 1993, p12). Medical classifications are seen as not merely describing the world but also modelling it, 'producing favoured readings of the body and the world at large' (Bowker and Star, 2000, p102). Underlying these readings of the body are the social and political forces that came to shape them. Thus how to describe disease and disability, what categories to include and what categories to exclude, are all actions influenced by social and political factors.

In this way Bowker and Star's much cited work on medical classification systems resonates with recent work in the sociology of diagnosis. Their work is highly relevant to the sociology of diagnosis, particularly as to date much of the focus of the sociology of diagnosis has been on diagnosis as category and the social and political forces at play in the construction of medical categories. Jutel asserts that category becomes, rather than is (Jutel, 2011a) and diagnosis cannot be isolated from human agency and application (Jutel, 2011a, p39). How the nosological boundaries are managed, the splitting and grouping of conditions into like or different, depends on many social factors and may be shaped by different stakeholders. As social constructs, medical classification systems are in perpetual flux as notions of disease and disorder change across time and culture and as different interests are served. They are not a map of a world of disease and disorder that exists ontologically without discussion, dispute or interpretation. As Armstrong suggests, there is no vantage point from which the truth of a pathological condition can be objectively seen as disease classifications are ever changing.

When classificatory systems and explanatory frameworks are in flux there is no Archimedean point from which to see things as they really are (Armstrong, 2011, p806)

The study of how diagnoses come to be or cease to be is a valuable illustration of how cultural, social and political forces shape the diagnoses available to any one society at any one time. There is an abundance of theorising about medicalisation, both historically from the 1960s onwards and in contemporary scholarly work including sociology of diagnosis publications. Much of this work is fascinating and highly relevant to the study of diagnosis as category. Since exploring this literature, I have learned much about how diagnoses (in the DSM in particular) come to exist, and what or who drives the creation of new diagnostic categories. While these debates are interesting and reveal much about categories of diagnoses, they are not as directly relevant to the experience of living without a diagnosis for parents of disabled children as other work in this field. The parents in my own study had children with accepted disabilities. Other than the families highlighted as having atypical experience, all of the children had difficulties that were mutually agreed by parent and practitioner to be medical in nature. The body of literature that examines contested diagnoses and medicalisation, including cultural influences on diagnostic categories and the growth of medicalised conditions, can only go so far in providing a theoretical framework for understanding the experience of living with accepted disabilities that have no specific diagnosis. For this reason, here I briefly consider the increasing medicalisation of conditions of everyday living and acknowledge that stakeholders may influence categories of diagnosis, but I do not go into in-depth detail about this work given that its relevance to my topic is limited. It is important to acknowledge the contribution of this literature, however, to make sense of our contemporary categorical, symptom-based system of diagnosis and our contemporary diagnosis-driven culture.

Much of the work of medicalisation considers psychiatric diagnoses, and indeed the roots of the sociology of diagnosis also lie in the study of psychiatric conditions (Brown, 1995). In psychiatry, dependence on standardised diagnosis is relatively recent (Godderis, 2011) and we have moved from the crude classification of 'idiocy' and 'insanity' in the mid 19<sup>th</sup> century to the multiplex system of the *Diagnostic and Statistical Manual of Mental Disorders* (DSM) today (DSM V, 2013). The third revision of the DSM in 1980 saw significant change in the way the classification system was organised, with the DSM-III (1980) revamped in line with

a classificatory biomedical approach to mental illness driven by a Neo-Kraepelinian heritage. The publication of the DSM-III represented a fundamental paradigm shift from dynamic to diagnostic models of psychiatric thought. Godderis describes this as 'a paradigm shift from dependence on psychodynamic approaches to a medically inspired diagnostic model', that shifted diagnosis from a marginal role to the cornerstone of the framework of psychiatry and mimicked the taxonomical nature of the natural sciences (Godderis, 2011, p134). This shift represented a transformation to a categorical, symptom-based system of diagnosis (Kokanovik, Bendelow and Philip, 2012).

Medicalisation has been the subject of sociological study across four decades and scholars have often focused on how nonmedical problems become medicalised. Early medicalisation models tended to emphasise top down social control function of medicine (Zola, 1972; Friedson, 1970; Illich, 1976), while more contemporary models of medicalisation consider a wider range of stakeholders in the medicalisation process (Atkinson, 1995) and recent work in the sociology of diagnosis has accelerated and augmented our understandings of the 'engines of diagnosis' (Jutel, 2011a; McGann and Hutson, 2011) and the 'disease mongering' that may benefit the commercial interests of stakeholders (Payer, 1992). The pharmaceutical industry, for example, is increasingly seen to be an active party in the diagnostic process and an engine of diagnosis (Jutel, 2009; Jutel, 2011a); trading on the mutability of diagnosis (Lakoff, 2000, 2006) through the creation and expansion of new disease categories by the drug companies who are also developing the treatments (Ebeling, 2011). Medicalisation in its contemporary context sees a number of new players invested in promoting medical explanations for a range of conditions which may have previously been accepted as problems of living (Szasz, 1961) and an increasing reliance on biomedical explanations and pharmacological treatments as a fast and efficient way of dealing with characteristics previously held to be within the realms of the normal (Scott, 2006).

A trend in the increasing medicalisation of conditions previously seen as within the realms of normal is reflected in the decisions made about which disease categories are included in each new edition of the DSM. Many of the new conditions in each subsequent DSM (beyond 1980) were previously seen as normal, if deviant, behaviour (Kokanovik, Bendelow and Philip, 2012) and there is significant body of work examining conditions that may previously have been accepted as a normal part of living like sleep difficulties (Coveney, Nerlich, and

Martin, 2009), shyness (Scott, 2006) and sadness and depression, (Horwitz, 2007, 2011; McPherson and Armstrong 2006). Ahuja (2003) has chronicled the increasing tendency of the 'worried well' to consult clinicians about emotional issues like financial stress, mourning, and shyness. There is also a large body of work examining the diagnosis and treatment of attentional difficulties in children (McKenzie and Wurr, 2004), both prior to and included in the work of the new sociology of diagnosis. This work includes consideration of the way that children from different cultures are assessed for ADHD and the potential role of culture in classification systems (Vallee, 2011); the growth of medicalised categories like ADHD (Bringewatt, 2011); clinician reservations about the diagnostic validity of ADHD as it is described in the DSM IV (Rafalovich, 2005); how laypersons and professionals navigate challenges to the legitimacy of ADHD (Fuller, 2011); whether Asperger Syndrome is a disorder or a neurological difference that has been socially constructed as a disorder (Molloy and Vasil, 2002) and the vanishing disorder of Asperger's (Singh, 2011). In this work on medicalisation and diagnosis as category, how the normal is differentiated from the pathological is considered, as well as cultural influences on diagnostic categories, and the shifting boundaries of normative expectations of contemporary Western culture.

Another site of dispute in diagnosis, well recognised by medical sociologists in the past and within the sociology of diagnosis, are conditions described as contested diagnoses and medically unexplained symptoms (MUS). The expression 'medically unexplained symptoms' has gained currency over the last three decades (Greco, 2012) and is now used routinely in clinical literature (Nettleton, 2006). Estimates of prevalence vary greatly as there is no consensus on how unexplained symptoms should be diagnosed, categorised and named (McFarlane et al., 2008), although Kokanovik, Bendelow and Philip (2012) suggest that 25-50% of primary care physician consultations in the UK are for dealing with difficult to define bodily complaints resulting from emotional or psychological issues with medically unexplained symptoms. In Western countries an increasing number of people report physical pain that cannot be medically diagnosed and people with MUS are held to be deviant cases for whom a classification rule cannot be applied (Mik-Meyer and Obling, 2012). The conditions considered contested and unexplained (for example fibromyalgia, irritable bowel syndrome, and chronic fatigue syndrome) have little in common and the patient group is not homogenous (Mik-Meyer and Obling, 2012) thus MUS is a container category. It contains individual symptoms of patients and also works as a reductionist label



that organises different patients into a group that has the illusion of unity; an unintended consequence of this classification is that the label of MUS label produces, reifies and stabilises the 'diagnosis' of MUS (Mik-Meyer and Obling, 2012).

Yet MUS is not a diagnosis in that it does not have the *meaning* of a diagnosis. Greco (2012) describes the term MUS as a 'polemical knot' (2012, p4), a 'placeholder: as a noun that works as a pronoun (eg 'thingamijig')' (2012, p2), in the sense that it's referent is not fixed to a single concept but alters according to context, taking on different meanings in different contexts.

MUS is not itself a diagnosis but rather a diagnostic no man's land; it does not perform any of the positive functions diagnoses are meant to perform (Greco, 2012, p2)

Nettleton (2006) suggests that people who live with MUS experience 'embodied doubt', an absence of truth; 'The 'truth' of MUS is an absence of 'truth'' (2006, p1176).

Classification systems, like the ICD or the DSM, have a way of dealing with conditions that are difficult to classify. Like the term *medically unexplained symptoms*, however, the residual categories of such classification systems may not be meaningful to patients or families. Furthermore work in medical sociology, including in the sociology of diagnosis, has examined the increasing cultural drive to diagnose and the increasing medicalisation of 'ordinary life' (Frances, 2013). For parents of children with significant, life-limiting disability yet no diagnosis, this cultural trend may be incongruous to their experience.

While sociological work on contested or medically unexplained conditions is valuable for my own topic in emphasising the significance of diagnosis for legitimising illness and for the function of creating meaning (Nettleton 2006, Madden and Sim, 2006), uncertain diagnosis in the context of my own empirical research is essentially different to contested diagnosis and medically unexplained symptoms. The children's difficulties in my research are held by both family and doctors to have organic cause; their status as medical conditions is not in dispute. In this way their undiagnosed disability does not fit with Dumit's characteristics of contested diagnoses (Dumit, 2006), nor Brown's four categories of diagnoses ranging from routine, unproblematic definitions through to intensely contested definitions (Brown, 1995).

*Absent diagnosis for uncontested medical conditions* may be seen as a new area of diagnostic tension; new in that it is currently under-theorised and that there are gaps in empirical work looking at the lived experience of negotiating absent diagnosis in this context. Thus while the existing theoretical literature in this area is relevant to the topic, it does not go far enough in considering absent diagnosis in the context of accepted childhood disability.

### *The sociology of diagnosis*

So far I have considered several interconnected concepts and areas of literature relating to diagnosis and its history: differentiating, the beginnings of diagnosis, the notion of the norm, and classification and medicalisation. Now, I turn to look at the newly emergent sociology of diagnosis. I have so far considered authors from key sociology of diagnosis publications when reviewing the themes above. Here, I look more specifically at the sociology of diagnosis as a sub-field and consider its relevance to my topic.

Despite the importance of diagnosis in medical theory and practice, diagnosis has not previously had its own sociology. The study of diagnosis has been, until very recent years, considered under the rubric of other subjects in medical sociology and in the sociology of health and illness. There has been an absence of the sociological study of diagnosis, as its study has been subsumed under other headings (Anspach, 2011). There is considerable literature around the subject of diagnosis and an abundance of sociological analysis underpinning it (Armstrong, 2011) so it has not been disregarded by medical sociology. Diagnosis has, for example, been considered by scholars writing about medicalisation, the sociology of science and technology, lay epidemiology, disease theory and the history of disease, classification and its consequences, contested diagnoses and medically unexplained symptoms, labeling and deviance theory, and the doctor-patient relationship. There are studies of diagnoses and their consequences, and of the diagnosis moment in the context of illness and disability experience and narratives. Studies that have been made of diagnosis have often been studies of diagnoses, one label in isolation from another (Jutel, 2011a). Diagnosis has not been ignored, but until recent years there has been no study of diagnosis

as a discrete subfield of medical sociology deserving of the title 'a sociology of diagnosis'. As Jutel writes:

The sociology of diagnosis does not have a clear identity or literature, hanging more on to the coat tails of medicalisation, disease theory and history of disease. It's not that diagnosis has been excluded from medical sociology, it's simply that it has been well buried in these and other areas of focus, and whilst pivotal, it hasn't been clearly isolated from these interests (2009, p279)

In this way diagnosis has been described as having an absent presence in the sociology of health and illness (Jutel and Nettleton, 2011).

The suggestion from within sociology that diagnosis merits more comprehensive study reaches back to the 1970s. Blaxter (1978) in her paper 'Diagnosis as category and process: The case of alcoholism' states that 'the activity known as diagnosis is central to the practice of medicine but it is studied less than its importance warrants' (Blaxter, 1978, p9). In this paper, Blaxter looks at the new diagnosis of the disease of alcoholism. She introduces the idea of diagnosis-as-category and diagnosis-as-process. She then examines diagnosis-as-category in the context of medical classification and diagnosis-as-process in the context of what the doctor is doing when he uses categories to make a diagnosis, proposing a theory of diagnosis as art and science. In highlighting the case of alcoholism she examines its inclusion as a disease in the ICD and the way that it is diagnosed, to assess how social diagnoses like alcoholism fit into doctors' normal classificatory systems and their practices. In doing so, she identifies a discrepancy between the inclusion of this 'social diagnosis' in the medical classification system of the ICD and the reluctance of doctors to make use of this diagnosis. Blaxter's work on diagnosis is commonly cited in contemporary sociology of diagnosis literature and, indeed, her categories of diagnosis-as-process and diagnosis-as-category are used to organize and structure the work on diagnosis by key sociology of diagnosis authors (Jutel, 2009; Jutel and Nettleton, 2011; Jutel, 2011a).

More than a decade after Blaxter (1978) proposed that the activity known as diagnosis was less studied than its importance warranted, Brown (1990, 1995) made a clear call for a more comprehensive focus on the study of diagnosis, coining the phrase 'sociology of diagnosis' (Brown, 1990). Brown (1990) in his paper 'The Name Game: Toward a Sociology of Diagnosis', like Blaxter (1978), emphasises the importance of diagnosis. His focus is

primarily on the theory and practice of psychiatry and psychiatric diagnosis. Describing the field of the sociology of diagnosis as 'disconnected' and still new, Brown states that diagnosis is 'loosely studied' and that social scientists have not yet cultivated a 'comprehensive approach' to diagnosis. He seeks to present an outline for a sociology of diagnosis, in which he focuses on psychiatric diagnosis but suggests many of the issues can be extended to medical diagnosis. While his later work has more of an emphasis on controversial and conflictual diagnoses, nowhere in his work is attention given to conditions that are agreed upon as medical but for which there is no diagnostic label. As his work focuses so precisely on psychiatry, and excludes consideration of disability, it was difficult to synthesise with my own topic. At the beginning of my journey of reviewing relevant literature I was hopeful when I discovered Brown's work as it further confirmed the importance of the sociological study of diagnosis and promised the beginnings of a sociology of diagnosis as a sub-field of medical sociology. Yet little of the analysis of diagnosis offered by Brown (1990, 1995, 2011) was relevant to my own study beyond the general drive to highlight the social construction of disease and how diagnosis is a relational process, and his exploration of the process of discovering and labelling conditions.

Brown's call for a sociology of diagnosis (Brown 1990, 1995) was not comprehensively answered until 2009. Annemarie Jutel (2009, 2011a, 2011b, 2014) has done much towards building this sociology of diagnosis and her work in recent years may be seen to mark a defining moment in which a new subfield of medical sociology has emerged (Anspach, 2011, pxiii). Jutel (2009) identifies her article 'Sociology of diagnosis: a preliminary review' as a first step in the drawing together of a number of threads in medical sociology that can contribute to this emerging sociology of diagnosis. This article was received within medical sociology as a touchstone essay seen to provide 'a roadmap of recent literature surrounding the terrain of diagnosis' (Hutson, 2011, xxxii). Following this article, three key publications in 2011 augmented the sociology of diagnosis as a new sub-field of medical sociology, these were: Jutel's book *Putting a Name to it: Diagnosis in contemporary society* (Jutel, 2011a); the collection of papers published in the edited book *Sociology of Diagnosis* Volume 12 Advances in Medical Sociology (edited by P J McGann and D J Hutson, 2011); and the collection of papers published in the special edition on the Sociology of Diagnosis in *Social Science and Medicine* 73 (2011). Jutel and Nettleton co-author the introduction to this special edition (Jutel and Nettleton, 2011). As David Hutson describes in his introduction to *Sociology of Diagnosis*

Volume 12 *Advances in Medical Sociology* (edited by P J McGann and D J Hutson, 2011); 'after laying dormant for years, the field of diagnosis grew exponentially within a very short period' (Hutson, 2011, xxx). The rapid augmentation of work contributing to this emerging field has been described as a 'diagnostic turn' in sociology and related disciplines (McGann, 2011).

The aggregate work on the sociology of diagnosis decisively establishes the importance of diagnosis and the need for a sociology of diagnosis. The key sociology of diagnosis authors highlight both the absence of a clear sociology of diagnosis, despite calls for its establishment, and the importance of diagnosis (Brown, 1990, 1995; Jutel, 2009, 2011a, 2011b, 2014; Nettleton and Jutel, 2011). Receiving a diagnosis is described by Jutel (2011a) as analogous to being handed a road map in the middle of a forest.

It shows the way-but not necessarily the way out. It indicates what the path ahead is going to look like, where it will lead, the difficulty of the climb, and various potential turn offs along the way. Perhaps it identifies the destination, but not necessarily. With a diagnosis, things don't necessarily get better, but they become clearer. The unexplained becomes explained, and management is defined. Assumptions are made about the future. The diagnostic moment is simultaneously transformative and contingent (Jutel, 2011a, p1)

The utterance of the diagnosis is played by Jutel as marking a boundary, serving to divide life into the 'before' and 'after'. The purpose of diagnosis is described by Jutel in her preliminary work (2009) as being multiplex, as: organizing illness, aiding administrative work, enabling access to services and status, giving permission to be ill and granting exemption from normal roles, guiding medical care, and reflecting what society is prepared to accept as normal and what should be treated (Jutel, 2009). Jutel proposes that medicine's authority is embedded in diagnosis at the institutional and individual level in the ability to assign categorical status and to define and delimit behaviours and people (Jutel, 2009).

The structure of the publications laying out a sociology of diagnosis is purposeful. Blaxter's dual definition of diagnosis as diagnosis-as-category and diagnosis-as-process is referred to frequently in the sociology of diagnosis literature. Further, in the recent key publications delineating the new sociology (Jutel, 2011a, 2014; Jutel and Nettleton, 2011; McGann and Hutson, 2011) the writing about diagnosis has been structured to reflect the dual definition

of diagnosis outlined by Blaxter, with a third consideration of diagnosis-as-consequence added in creating a three part structure for the sociological study of diagnosis. Although Jutel (2009) resisted structuring her paper based on the division of diagnosis into process and category, recognizing the two as 'intertwined, making it difficult to speak of one without considering the other' (2009, xiv), Jutel and Nettleton (2011) categorise later work in the sociology of diagnosis in terms of process, category and consequence. They are self-reflexive about this, describing their power as editors to classify their subject and to construct categories, citing Thomas Arnold (1839) when they say 'it will divide wherever we choose to strike it'. Their decision to 'coax the papers in this special issue into these fields' (Jutel and Nettleton, 2011, p798) excludes other possible analytic approaches, which they acknowledge. It is suggested that, like the chicken-and-egg situation, the creation of diagnosis and their consequences is not a one-way model. The social consequences of diagnosis mold the disease classifications, as do the classifications frame the experience of diagnosis (Jutel, 2011a, p143). Diagnosis-as-process and diagnosis-as-category depend on each other and the relationship is described by Jutel as circular.

There are multiple distinct themes examined by these key publications that established the new sociology of diagnosis. Inevitably these themes overlap and intertwine and many of the themes are not new to medical sociology. But what is unique about the sociology of diagnosis is that for the first time these areas of interest in the study of diagnosis are brought together and examined with the aim of achieving a more comprehensive sociological examination of diagnosis. The areas examined by this work include: medicalisation; classification; areas of tension in diagnosis with reference to the illness-disease distinction, contested diagnoses and medically unexplained symptoms, and non-medical agents of diagnosis; the doctor-patient relationship; illness narratives; lay epidemiology; and the technologies of diagnosis and the sociology of science and technology.

Jutel (2011a) proposes that future directions for the sociology of diagnosis field must be concerned with the *creation, application, allocation* and *exploitation* of diagnostic categories. Jutel and Nettleton (2011) also highlight the issues and agendas for a future sociology of diagnosis, stressing the important of exploring 'the way classification and labels are constructed, framed and enacted' (2011, p798). Anspach (2011) lays stress on the importance of considering diagnosis in the context of the *practice* of medicine in a sociology of diagnosis.

She states, 'a sociology of diagnosis demands that we examine the full array of sites in which a diagnosis is practiced' (xxiv). Hutson (2011), in the same volume, proposes a sociology of diagnosis considers: the role of diagnosis, the power of categories, nosological authority and the meanings attached to diagnoses of health and illness. Their emphasis for the future of the sociology is on diagnosis as both category and process.

The structure of the sociology of diagnosis is thus suggested by several scholars, principally Jutel. However, despite recommending the study of diagnosis as process, category and consequence, in Jutel's work, diagnosis as category is the dominant focus (2009, 2011a, 2011b) In her exploration of medical classification in her book length work on the sociology of diagnosis (2011a) she examines the history of diagnosis and medical classification systems; the social framing of diagnosis via analysis of corpulence and fetal death, which she has previously written extensively about; contested diagnosis and medically unexplained symptoms; and the 'peddlars and pushers' (Jutel, 2011a, p97) who drive diagnoses and the multiple engines of medicalisation and the commercial interests served with disease mongering. The chapter of the publication most relevant to diagnosis as process is that looking at diagnosis and the doctor-patient relationship. Here, illness narratives are acknowledged and discussion on self-diagnosis explores the changing roles in diagnosis. Self-diagnosis has been the subject of Jutel's other work both before and after the publication of the key sociology of diagnosis texts.

Similarly in McGann and Hutson's (2011) volume *Sociology of Diagnosis*, the principal contribution is to the study of diagnosis as category. Ten of the 14 papers included in the volume could be described as having diagnosis as category as their principal focus. Only one article in the volume looks specifically at the process of diagnosis, and in doing so examines psychiatric diagnosis (Godderis, 2011). While all articles have elements of the multiplex meanings of diagnosis (as process, as category and as consequence) there is substantial focus on diagnosis as category, and in particular on psychiatric diagnosis as category, and very little focus on the process of diagnosis. Other authors introducing the sociology of diagnosis offer a more robust examination of diagnosis as process. In the sociology of diagnosis special edition of *Social Science and Medicine* (2011), there was a stronger presence of papers looking at diagnosis as process, and several reporting ethnographic studies of the process of diagnosis.

I suggest that in the key sociology of diagnosis literature to date, there has been insufficient work on diagnosis as process. The sociology of diagnosis has been successful in examining diagnosis as category and through this work we now have a much greater understanding of the multiplex dimensions of the classification and medicalisation of diagnosis, including the drivers, engines, and peddlars and pushers of diagnosis. How a diagnostic category comes to be, particularly with reference to contested conditions, has been deconstructed in this and other work (Bowker and Star, 2000). Yet, if the field wants to move away from the 'coattails of medicalisation' (Jutel, 2011a, p6) then it needs to more comprehensively examine and consider diagnosis as process and to do so in 'the full array of sites in which it is practiced' (Anspach, 2011, xxiv). This could include a more comprehensive examination of how the patient, or parent of patient, may be active in the diagnosis process and negotiate or co-construct diagnosis; ethnographic work on the process of diagnosis would be pertinent here. A consideration of the process of diagnosis as a hermeneutical enterprise (Leder, 1990), involving multiple players and texts, is partly theorized in the sociology of diagnosis work but receives little empirical attention and remains under-theorised. The disproportionate attention to diagnosis as category can perhaps explain why Jutel in her key texts (Jutel, 2009, 2011a, 2011b) excludes the work of eminent sociologists relevant to the study of diagnosis as process, including Armstrong, as well as writers in other fields examining the process of diagnosis in work that was published prior to the key sociology of diagnosis publications (for example Featherstone et al., 2005; Featherstone et al., 2006; Featherstone, Gregory and Atkinson 2006; Latimer 2007a, 2007b; Latimer et al., 2006; McLaughlin 2005; Shaw et al., 2003). Increased focus on how diagnosis is *done* would create a more equal balance of interests, which currently is weighted towards the consideration of how diagnosis categories are *created*.

The sociology of diagnosis could also expand by considering its dialogue with genetic diagnosis and I suggest genetic technologies and advances in genetic knowledge are not given sufficient weight as 'drivers' of diagnosis in the sociology of diagnosis work to date. Although the input of technology in the creation of new diagnoses and new genetic technologies are referred to, there is simply not enough consideration of the impact of genetic diagnosis. Increasingly, genetic explanations are considered for a whole plethora of disorders, from alcoholism to unexplained development delay, yet in sociology of diagnosis



work more attention is given to the role of the pharmaceutical industry in the creation of diagnoses than the expanding realm of medical genetics. Certainly, the role of big pharma as a 'peddler or pusher' of diagnosis brings with it scandal and story, but there is also a growing body of work considering the process of genetic diagnosis, particularly for childhood disorders, and other important aspects of dysmorphology (Dimond, 2014a; Featherstone et al., 2005; Featherstone et al., 2006; Featherstone et al., 2007; Featherstone and Atkinson, 2013; Latimer, 2007a, 2007b, 2014; Latimer et al., 2006; McLaughlin, 2005; McLaughlin and Clavering, 2011; McLaughlin and Clavering, 2012; McLaughlin, 2014). While a number of these journal articles and monographs were published after the key sociology of diagnosis publications in 2009 and 2011, those published prior to 2009 were not discussed in work laying down the new subfield of medical sociology. The sociology of diagnosis would be enriched by the inclusion of this and later work examining the process of genetic diagnosis and this would improve the dialogue with both the process of diagnosis and genetics.

In terms of specific relevance to my topic, the sociology of diagnosis does not consider conditions that are agreed upon as medical but for which diagnosis is absent. I suggested above that absent diagnosis in the context of my topic represents a new area of diagnostic tension; that is, uncontested medical conditions without diagnosis. The work considered as forming the new sociology of diagnosis lacks consideration of long-term physical disability, instead focusing primarily on psychiatric labels and contested or disputed diagnoses. There are often controversial definitional, political, financial or social control battles behind the scenes of the diagnoses that are examined in this work. Childhood disability does not come with such sensationalism, and absent diagnosis is not potentially attributable to political or financial motives. But the story this thesis will tell is one of the *meaning* of diagnosis for parents. Absent diagnosis can tell us much about diagnosis, and this is not currently considered by the sociology of diagnosis. The refraction of diagnosis, its absence, shines a light on what diagnosis is and means. In the natural breaching experiment (Garfinkel, 1963) that is living without a diagnosis, the perceived meaning and multiple purposes of diagnosis is exposed. To find the nucleus of diagnosis, we must consider its absence as well as its presence; for boundary work involves knowing what is on either side of the line. The way that a patient or parent deals with uncertainty or absence of diagnosis can be telling of what

we expect from diagnosis, yet the sociology of diagnosis does not as yet include the empirical or theoretical study of this topic in this context.

### 2.3 The process of paediatric genetic diagnosis

As suggested, the current focus of the sociology of diagnosis does not give sufficient consideration to the process of diagnosis. In particular, adequate consideration is not given to the process of genetic diagnosis in the current sociology of diagnosis work. The genetics clinic is one of the key sites in which diagnosis is practiced, increasingly so for the diagnosis of childhood developmental disorders. In the UK, children with developmental difficulties are increasingly likely to be referred for genetic investigation as part of the exploration of what is causing their difficulties (Department of Health, 2003 cited by McLaughlin and Clavering, 2011). McLaughlin and Clavering (2011) report that the rise in referrals is related to a number of factors, including the increasing precision of new genetic testing techniques.

There is a growing body of research within sociology and anthropology exploring diagnosis in paediatric genetics (Dimond, 2014a; Featherstone et al., 2005; Featherstone et al., 2006; Featherstone, Gregory and Atkinson, 2006; Featherstone and Atkinson, 2013; Latimer, 2007a, 2007b, 2014; Latimer et al., 2006; McLaughlin, 2005; McLaughlin and Clavering, 2011; McLaughlin and Clavering, 2012; McLaughlin, 2014; Shaw et al., 2003). This work is primarily ethnographic and is highly relevant to the study of diagnosis and forms an important foundation from which to explore the diagnosis of childhood disability, particularly in light of recent advances in genetic testing techniques available to families in the UK.

This work deconstructs the process of paediatric genetic diagnosis, exposing key conceptualisations that underpin this process, including the uncertainty, undecideability and instability of diagnosis in genetics and how diagnosis is played as deferred (Latimer et al., 2006; Latimer, 2007a, 2007b, 2013) and how clinical authority is exercised within consultations with families (Featherstone et al., 2005). In this body of work, there is a focus on parental participation in the diagnosis process (see Dimond, 2014a; Latimer, 2007a, 2007b, 2013 and also McLaughlin, 2005; McLaughlin and Clavering, 2012; McLaughlin, 2014) and

the visual culture of dysmorphology (McLaughlin and Clavering, 2012; Shaw et al., 2003; Featherstone et al., 2005; Latimer et al., 2006; Latimer, 2013; McLaughlin, 2014). Work in this field also considers the moral and sentimental work of the clinic (Featherstone, Gregory and Atkinson, 2006) and issues relating to kinship and responsibility (McLaughlin and Clavering, 2011; Featherstone et al., 2006).

In the years spanning my study, there have been profound changes in knowledge in medical genetics and tests available to diagnose genetic disorders. One example of the new genetic technologies available now, that was not available for clinical or research purposes at the beginning of my study in 2005, is the ability to carry out exome sequencing in a research capacity for children with unexplained developmental disorder. One of the aspirations of the Deciphering Developmental Delays (DDD) study is that this technique of sequencing the exome of affected children and their parents to identify existing and new gene mutations is rolled out to the NHS for all relevant families. It is anticipated that when the DDD study stop recruiting families in 2015, that the clinical work of looking for a diagnosis via exome sequencing will be carried on by the NHS. This represents a radical change in the way genetic testing is done. The recognition that unexplained disorders of childhood may have genetic aetiology is a powerful aspect of our current thinking about childhood disability.

The identification of conditions as genetic is a significant feature of current medical thought and perception (Featherstone and Atkinson, 2013, p12)

There is now a new level of variation able to be seen and diagnoses made that were not previously possible (McLaughlin, 2014). In the four decades since the first genetic investigations, genetic science has advanced expeditiously, and now much smaller molecular and chromosomal changes can be determined (Featherstone et al., 2005). Increasing use of DNA microarray analysis (Featherstone and Atkinson, 2013) further enhances the ability to locate specific genetic diagnosis. The impact of new genetic technologies on the work of the clinic and the consequential shift in the process of diagnosis has been described both as the 'death of the clinic' (Haraway, 1991) and its 'rebirth' (Latimer et al., 2006; Latimer, 2013). The profound rate of growth in new genetic techniques and the identification of a genetic aetiology for an increasing range of conditions and disorders has substantial implications for medicine and important ethical, legal and social implications.

Authors writing about the process of paediatric genetic diagnosis in the context of unexplained childhood disability define their work with reference to the discipline of dysmorphology, with some key authors (for example, Featherstone and Latimer) more explicitly contextualising their work with relation to dysmorphology than others (like McLaughlin and Clavering) who make reference to dysmorphology but describe their work as taking place in the context of paediatric clinical genetics. Here, I will use both terms interchangeably. Shaw et al., (2003) highlight the dual focus of the medical speciality of clinical genetics as being the risk assessments in relation to developing conditions such as breast cancer, and the diagnosis of conditions involving dysmorphic or unusual forms. Dysmorphology is thus seen as a sub-speciality of clinical genetics. Dysmorphology is 'the professional discipline of delineating disorders affecting the physical development of the infant, before or after birth' (Featherstone et al., 2005 p11). Latimer (2013) highlights how dysmorphology has been 'at the heart' of the emergence of medical genetics in the UK (2013, p54) and suggests an expanding interest in the delineating of congenital abnormalities or syndromes fuelled by developing technologies to 'visualise' the chromosomes and genes (2013, p55). Medical genetics is a relatively new discipline, around 50 years old: its early association with a eugenic ethos and practices potentially curtailed its progress as a discipline (Latimer, 2013, p52).

Dysmorphology work involves families, not just individual patients. McLaughlin and Clavering (2011) argue that even when paediatric genetics finds no genetic explanation for a child's difficulties, or suggest a de novo mutation, questions around kinship are still significant and kinship is unsettled. Whether paediatric genetics produces new or different understandings of kinship relations (Featherstone et al., 2006) is one of the considerations of this primarily ethnographic work looking at diagnosis in dysmorphology. This work looks specifically at how genetic categorisation is accomplished (Latimer et al., 2006). The process of categorising paediatric patients involves surveying patterns of physical features in the child and their family. Through a process of alignment and exclusion, pathology becomes visible as the body's signs and symptoms are read and clusters of associated features and symptoms identified (Latimer et al., 2006).

This work has identified that in the field of dysmorphology it is often difficult to give a definitive genetic diagnosis (Latimer et al., 2006). While some parents received a diagnosis of

a named syndrome for their child relatively quickly once they had been referred to the clinical genetics service, for the majority the process of attendance at the clinic and the search for a diagnosis continues over years (Featherstone et al., 2006). Deferral of diagnosis, and 'keeping people on' at the clinic (Latimer et al., 2006, p661) may characterise the process of genetic diagnosis in this context. For many syndromes, a mutation at the molecular level has not yet been identified, despite the 'extraordinary disruption' (Latimer, 2013) that many children experience in their development.

Although most dysmorphic syndromes are thought to involve sub-microscopic genetic mutations, and changes in DNA sequence, for many syndromes there are not yet any specific tests (Latimer, 2013, p89).

Beyond the work described here, there has been little focus on either the process of genetic diagnosis in this context or what happens when diagnosis is not achieved.

Many studies on genetic medicine focus on the impact of a genetic diagnosis, but do not particularly attend to how, and at what moments, a genetic diagnosis is accomplished. Nor indeed, and just as importantly, do these studies note when it is not accomplished (Latimer, 2013, p103)

The relevance of ethnographic studies of dysmorphology for my topic is the attention to the 'messy' work of classification (Bowker and Star, 2000) that takes place in and beyond the clinic, including parental participation in that work, and the method of doubt and deferral upon which the process rests. Parents are immersed in this clinical space of deferral, shifted between the experience of definition and uncertainty (Latimer, 2007a). Featherstone and Atkinson encourage us to look 'backstage to the many sites where the syndrome is enacted' (2007, p213). The categorisation of syndromes is not static and the classification and description of genetic syndromes are subject to change, with some conditions representing nosography-in-the-making (Featherstone et al., 2005). In examining the process of paediatric genetic diagnosis we have the opportunity to observe the process of genetic classification as it occurs, and also how that process is enacted by both clinicians and families.

Paediatric genetics involves multiple visually based diagnostic processes (McLaughlin, 2014) and the visual culture of dysmorphology is described by a number of authors writing about the process of paediatric genetic diagnosis (McLaughlin and Clavering, 2012; Shaw et al., 2003; Featherstone et al., 2005; Latimer et al., 2006; Latimer, 2013; McLaughlin, 2014). The process of making a genetic diagnosis involves close examination of the child's body in order to identify clues to possible genetic mutation (McLaughlin and Clavering, 2012) and many syndromes are recognised as having a particular look or face (Latimer, 2013). In addition to the molecular testing process, family stories are gathered, as are photographs of the affected child and their family, in this process of looking for clues. Visual representation plays a central role in the creation and transmission of medical knowledge in this field, and the photograph is commonly used in the process of classification in the dysmorphology clinic with photographs read for signs of dysmorphia (Featherstone et al., 2005). The camera is thus the vehicle for categorising difference (McLaughlin and Clavering, 2012). In clinical genetics the photographic image of the individual patient, and the surveillance of his or her appearance, persists (Featherstone et al., 2005) and the clinic is described as 'a site for the spectacular display and representation of bodies, organs and pathologies' (Featherstone et al., 2005, p552). Visual representations of the child are routinely taken outside of the clinic, for example to local dysmorphology meetings, and may be presented nationally and internationally if the child's disorder appears interesting, borderline, subtle or difficult to classify (Featherstone et al., 2005).

A particular kind of gaze is germane to the dysmorphology clinic, and clinicians may be skilled in the art of seeing a syndrome, enacting the gestalt of dysmorphology (Latimer, 2013). At times the recognition of a syndrome may be instantaneous, if the syndrome is clearly expressed or if a clinician is particularly skilled in the art of seeing that syndrome; other times further work is needed to locate (or defer) diagnosis. Dysmorphology provides a contemporary example of the clinical gaze in action (Shaw et al., 2003, p9 drawing on Foucault, 1973). The clinical expert, in this literature, is seen as having the power and ownership of the interpretation of the significance of the visual representations; 'oracular pronouncement' (Featherstone and Atkinson 2013, p.552) and the *craft* of seeing is

emphasised (Featherstone et al., 2005). The clinician decides what is normal from abnormal, familial from pathological; 'the clinician is the skilled reader and interpreter of visual difference and similarity' (Latimer et al., 2006). The photograph may be dismissed in favour of the ability of the expert to see a syndrome, or a molecular test may not rule out diagnosis if the expert clinician can see something in the patient that conflicts with this test result. In this way, craft expertise remains paramount to genetic diagnosis (Latimer, 2013) and in the contemporary process of paediatric genetic diagnosis there is a considerable role for expert judgement (Featherstone and Atkinson, 2013). While diagnosis and clinical classification are being transformed by genetic technologies, therefore, clinical judgement is still central, particularly as for many of the conditions encountered in the dysmorphology clinic no test is available (Featherstone et al., 2005). In the surveillance of photographic images, the gestalt or 'diagnostic intuition' of individual practitioners is a form of tacit knowledge that expresses an intuitive response to the visible features of a patient (Shaw et al., 2003). Shaw et al., (2003) report that some clinicians feel this skill depends upon an obsessive interest in classification and the recognition of patterns, an intuitive ability that may, ironically, be considered by dysmorphologists to be genetic (Latimer, 2013).

Visually based practices of diagnosis also take place outside of the clinic (McLaughlin and Clavering, 2012). McLaughlin and Clavering (2012) reported that parents and other family members followed up consultations with searches on the internet, becoming involved in the visual culture of dysmorphology as they surveyed images seeing differences or similarities between their own child and the children represented in images online. McLaughlin (2014) reports that it was not only common but expected that families reinforced or extended the information they received in consultations with time spent online outside of the clinic, researching the genetic syndromes and variations that had been discussed in the consultation. Parents accessed websites that mapped the features associated with particular syndromes and examined the faces and bodies of other children. The images they found had profound significance for parents. Making connection with others who look similar outside the biological family may be seen as a form of kinship making (McLaughlin, 2014, p12).

A key aspect of the process of genetic diagnosis in the context of childhood disorders is the role parents may play in the diagnosis process, in which they are not just recipients of diagnosis but participants (McLaughlin, 2005, p285). The work in this field identifies a transformation in the role parents play in the clinic (Dimond, 2014a). Previous analysis in medical sociology of the role of parents in the clinic suggested that professionals directed parental talk and behaviour in the clinic environment (Strong, 1979) and parents had a subservient role in the clinical encounter (Davis, 1982). However, as Dimond (2014a) illustrates, the clinical encounter has undergone significant change in recent years, with an increasing emphasis on shared decision-making and increasing use of the internet to access medical information and support. The literature reviewed in this section highlights the importance of parents in the paediatric genetic clinic, not just in accompanying their child to the clinic but as contributors, actors or partners in the genetic diagnosis process both inside and outside of the clinic (see in particular Dimond, 2014a; Latimer, 2007a, 2007b, 2013; and also McLaughlin, 2005; McLaughlin and Clavering, 2012; McLaughlin, 2014). The clinical team and parents can be jointly involved in a search for the right diagnostic category and parents become enrolled in the diagnostic process (Featherstone and Atkinson, 2013).

Latimer (2007a) suggests parental participation in the clinical work of diagnosis is remarkable and that they engage in the epistemological practices through which their children's problems gain definition (Latimer, 2007a). Parents are encouraged to participate in defining their child's clinical problems and use expert and techno-scientific discourses to do so, and so they are able to 'talk the talk of the clinic' (Latimer 2007a, p113). This is set in contrast to other medical settings where clinicians exclude patients and family members from the 'expert' work of diagnosis. This reflects findings by Brookes-Howell in her 2006 study on the interactional management of diagnostic uncertainty in the genetic counselling clinic in which she reports that, unlike other medical settings, clients within genetic counselling are able to actively contribute to discussions regarding diagnosis and for the extent of non-diagnosis to be negotiated. This research work builds on earlier studies that propose diagnosis as a product negotiated between practitioner and client, including the



work of Gill and Maynard (1995) who demonstrate that parents are able to take an active role in negotiating a diagnostic label for their child.

Especially for young children, the parental role of facilitator and narrator are a crucial part of the function of paediatric medicine (Dimond, 2014a). Parents and family members are made visible in the clinic and there is extension of the clinical gaze to include the parents' body and other close and extended family members as the clinician seeks to sort the normal from the abnormal, the familial from the pathological (Dimond, 2014a). In this way, parents become quasi-patients (Dimond, 2014a, p7). Dysmorphologists enlist parents in supplying photographs, in tracing a feature through the family, in the work of interpreting signs, symptoms and features, and in distinguishing what is familial from what is potentially syndromic (Latimer, 2013). As with the interpretation of visual representations of the child, the clinician retains authority to decide what is relevant from the knowledge shared by the parent. Therefore, although parents are expected to show interest and narrate their child's story, the significance of the knowledge is 'owned' by the clinician (Dimond, 2014a). The extent and way parents are involved is therefore contingent on the clinician's interpretation of what is significant or not. Parents are therefore both guide and guided (Dimond, 2014a) as clinicians either hold things in play or dispose of them (Latimer, 2013). McLaughlin (2005) suggests it is important to remain conscious of the 'embedded position' of medical professionals in constructing the processes through which parents enact with the clinic, potentially shaping the meanings developed in interactions (2005, p286).

Latimer also explores how the role of the clinic is not merely a diagnostic one, but it also has the major role of non-directive genetic counselling. The clinic essentially offers families not just diagnosis but information to assist with future reproductive decision making. The family, through this process, becomes a site of selection (Latimer, 2013). Latimer refers to Bauman (2003) in considering the selection opportunities families are faced with when receiving information about risk of recurrence in the clinic. Bauman (2003) suggests that with reproductive and genetic technologies people, as new consumers, have the tempting prospect of selecting their desired children: 'the chance to...choose a child from a catalogue' (Bauman, 2003, p40, cited in Latimer, 2013, p153). This represents a further dimension of how parents are involved in the clinic.

The binary attitudes of certainty and uncertainty are key themes in medical sociology (Atkinson, 1984, 1995; Fox, 1957) and uncertainty has been identified as a marked feature of genetic medicine (Latimer, 2007a, 2007b). In ethnographic work examining the process of genetic diagnosis, the dysmorphology clinic emerges as a space of deferral (Latimer et al., 2006; Latimer, 2007a, 2007b), with dysmorphologists performing genetics as a new frontier of knowledge and diagnosis as complex, uncertain and undecideable (Latimer, 2007b, p14). Parents are 'entangled' in this space of deferral (Latimer, 2007b, p21). In the field of dysmorphology it is often difficult to give a definitive genetic diagnosis; with the uncertainty in dysmorphology performed not as doubt but deferral (Latimer, 2007a, 2007b). Brookes-Howell (2006) advises that it is not uncommon for the client and genetic counsellor to be faced with uncertainty regarding diagnosis. This situation of 'non-diagnosis' (Brookes-Howell, 2006, p308) is described as one in which the client cannot be offered a diagnosis at a given point in time. Clients in the genetics clinic require certainty of diagnosis to prepare for the future, understand the present, and for peace of mind (Brookes-Howell, 2006, citing Skirton, 2001). Brookes-Howell (2006) suggests that these areas are difficult for parents of children faced with a non-diagnosis and that the uncertainty of non-diagnosis can result in confusion. Non-diagnosis can impact on the parenting role, creating a unique vulnerability and fear of the unknown that can disrupt parental adjustment (Brookes-Howell, 2006, citing Applebaum and Firestein, 1983). Brookes-Howell's work suggests that non-diagnosis is an important aspect of genetic counselling.

McLaughlin and Clavering (2012) highlight the significant uncertainty associated with identifying a new or complex genetic variation. Although there is a desire for certainty in the clinic, and indeed parents may make requests for unequivocal diagnosis from the geneticist (Featherstone and Atkinson, 2013), the classification of genetic diagnosis is 'as much about doubt, and the precarious nature of diagnosis, as it is about certainty' (Latimer, 2013, p8). There is therefore a tension between the existence of doubt and the desire for certainty. The science of medical genetics relies on the notion of deferral (Latimer, 2013) and 'keeping people on' (Latimer et al., 2006, p661) at the clinic is one way in which this deferral is enacted. The paediatric patient may be kept on at the genetics outpatient clinic because the

knowledge, rather than the child, may change or because the child is young and does not have the look of a particular syndrome but may still 'grow into the face of a syndrome' (Latimer, 2013, p104; Featherstone et al., 2005). This represents a commitment to the possibility of certainty and reflects an expectation that technologies will improve and more accurate or appropriate molecular tests will become available in the near future (Featherstone et al., 2005).

Parents are held to occupy a 'space of motility' (Latimer, 2007a, p126) in their participating in the clinic, shifting back and forth between deferral and definition as they negotiate potential diagnosis. The motility in which they are tangled (Latimer, 2013) is not held to be the same as liminality, for unlike Turner's (1987) liminoid space of sequestration, of being between two statuses (Latimer, 2007a, p113; Latimer, 2013) parents in the dysmorphology clinic are moved between these states through participation not sequestration or subjection. Again, this rests on the idea of there being a future path leading to diagnosis.

Parents are shifted back and forth between a space of definition (in which they help to show how their child's troubles belong to a category) and a space of deferral (in which that category is not yet fully known but promises a future of knowledge provided the right path is followed) (Latimer, 2007a, p113)

Studies like the Deciphering Developmental Delay study are examples of the way in which families come to the clinic for diagnosis and are enrolled as material for scientific investigation (Latimer, 2013). This happens because, in the absence of diagnosis, there is deferral. It is unclear what the impact on this space of deferral will be in the future exome or whole genome sequencing is offered as standard when a child presents with development delay. For now, in the absence of diagnosis, a child is kept on at the genetics clinic and diagnosis is deferred. Parents, like clinicians, may be committed to future scientific advances and the belief that advancement in science will provide more answers about their child's medical condition (Reiff et al., 2012). Families are enrolled as allies in the making of these futures (Latimer, 2013, p179) and the clinic gifts hope to the families; 'the gifts the clinic pass to the family are of a future, a future of definition, a future of classification' (Latimer, 2013, p397). In the meantime, families wait and see, a state of motility that Aase (1990) and McLaughlin and Clavering (2012) call watchful waiting.

Being kept on at the clinic may have other benefits for families. The clinic can be a site of reassurance (Latimer, 2013). Obtaining a genetic diagnosis and classification of their child's problems is not the sole function of the clinic, which is an important site for the sentimental and celebratory focus on the child (Featherstone, Gregory and Atkinson, 2006). The clinic is thus both a 'confessional space' where parental concerns about the aetiology of their child's condition can be discussed and feelings of blame potentially absolved, and also a sphere in which the development and behaviour of the child is discussed in favourable terms (Featherstone, Gregory and Atkinson, 2006). Parents are also acknowledged as experts on their child (Latimer, 2013).

Literature that examines the process of paediatric genetic diagnosis is relevant to my own topic. Many of the children of parent participants in my research were affected by conditions thought to be genetic and parents were involved in the process of seeking a diagnosis. Although it was not anticipated prior to the interview study, the themes identified in this work; of parental participation in the clinic, the visual culture of dysmorphology, and of the deferral of diagnosis with families kept on at the clinic, were relevant to the data generated in my interview study. This will be explored in the coming data chapters. The final section of this literature review considers research with families.

## **2.4 Living Without a Diagnosis: Research with Families**

As already suggested, there is a gap in the literature regarding research with families living without a diagnosis and in the sociological study of absent diagnosis. There is an established body of work on the experience of parenting a disabled child and the impact of caring on parents. This research literature, spanning the last three decades in particular, reports on the psychological, social, emotional and financial impact of caring for a disabled child or children. The implications, both socially and psychologically, of parenting a disabled child with a named condition are therefore well documented (Breslau, 1982; Gallagher, Phillips and Carroll, 2010). However, there are only a small number of studies looking specifically at the experience of parenting a disabled child without a diagnosis. This work is inconsistently conceptualised in that authors use a range of different terminology to define the context of their work. Terminology used to describe absent diagnosis in work

with families includes the following: 'undiagnosed families' (Genetic Alliance, 2010); 'undiagnosed medical conditions' (Madeo et al., 2006); 'an unknown diagnosis' (Graugaard and Skov, 2006); 'unidentified multiple congenital anomaly syndrome', 'MCAs', 'undiagnosed syndromes', 'without a definitive diagnosis', 'no unified diagnosis', 'absence of diagnosis', 'children with an unidentified syndrome', 'unexplained MCA syndrome' (Rosenthal, Biesecker and Biesecker, 2001); 'undiagnosed disorder', 'non-diagnosis', 'living without labels' (Brookes-Howell, 2006); 'non-diagnosed child', 'a child without a diagnosis', 'living without a diagnosis' (Lewis, Skirton and Jones, 2010). This lack of standardisation in the terminology to describe the topic can make for difficulties collating relevant literature. Clarification of the literature relevant to this topic is useful and no study to date has been reflexive about the inconsistencies in definitions in this field. Considering this literature as a body of work is timely given the recent emergence of the sociology of diagnosis, which heralds a commitment to drawing together a focus on social issues in diagnosis. Such a project of clarifying and establishing a coherent approach to diagnosis, seen in the diagnostic turn in medical sociology, would also benefit empirical work examining diagnosis.

Those studies looking directly at the experience of living without a diagnosis, or considering the perceived significance of diagnosis for families, are principally empirical studies utilising qualitative methods, most commonly interview studies with parents. Two studies are particularly relevant to my topic due to their focus exclusively on the perceived impact of absent diagnosis for families with undiagnosed disabled children (Rosenthal, Biesecker and Biesecker, 2001; Lewis, Skirton and Jones, 2010). Other studies are also relevant, however as suggested above they are not currently collated as a unified body of work. Some research work, for example, includes families with disabled children without a diagnosis by virtue of their sample having a number of families with undiagnosed children. The results of this work can be meaningful for investigating the lived experience of absent diagnosis, despite absent diagnosis not being the principal area of research interest. I identify the following literature as being most relevant to the study of living without a diagnosis for families with disabled children:

- A qualitative study with 29 parents of 16 children without a diagnosis recruited through a National Human Genome Research Institute in the United States. This

study looks at parental attitudes towards a diagnosis of children with multiple congenital anomaly syndromes (Rosenthal, Biesecker and Biesecker, 2001)

- A qualitative study with 14 parents of 9 children without a diagnosis recruited through a Regional Genetics Centre in the United Kingdom. This study looks at the parental experience of living without a diagnosis (Lewis, Skirton, and Jones, 2010)
- A longitudinal qualitative study of 16 parents of 8 severely disabled children in Denmark, half of whom did not have a diagnosis, recruited via a neuro-paediatric department, a neonatal ward and a department of clinical genetics. The authors look at the meaning of diagnosis and the experiences of parents of a severely disabled newborn child in relation to the diagnosis process (Graungaard & Skov, 2007)
- A qualitative study with parents of 20 children with Intellectual Disability, half of whom did not have a casual diagnosis, recruited through a genetics program in Canada. This study looks at the perceived value of a diagnosis and compares families with and without diagnosis (Makela et al., 2009)
- An electronically administered mixed method survey of 266 parents of a child, or children, affected by an undiagnosed medical condition across various countries in the developed world. Parents were recruited via disease advocacy organisations, patient support groups and online forums and the study looked at the factors associated with perceived uncertainty among parents of children with undiagnosed medical conditions (Madeo et al., 2012)
- A questionnaire study of 717 parents in Germany that assessed the psychological benefit of diagnostic certainty. This study compares mothers of non-disabled children to mothers of children with Down syndrome and mothers of children with diagnostically unassigned mental retardation (Lenhard et al., 2005)
- A mixed method study of parents and professionals carried out by the Genetic Alliance UK organisation. 149 parents took part in a mixed method questionnaire study administered online to parents recruited via the Syndrome Without a Name UK database; and 9 families and 6 health professionals/ charity staff in England took part in qualitative interviews. The study looked at the needs and issues faced by families with children affected by undiagnosed conditions (Genetic Alliance UK, 2010)
- An interpretive qualitative study with 14 parents of 13 families with children with developmental disability. Participants included 5 parents of children with

unidentified disabilities or nonspecific developmental delay. The study investigated the reasons why parents seek a differential diagnosis for their child and family experience of the diagnosis process (Watson, 2008)

This list is not exhaustive, and relevance is considered by degree rather than kind. Other work is relevant and is considered throughout this thesis; the literature noted above is felt to be the most relevant to the study of living without a diagnosis. Of this core literature, two studies (Rosenthal, Biesecker and Biesecker, 2001; Lewis, Skirton and Jones, 2010) are most relevant to my own research, both of which highlight the need for further work in this area. The earlier study by Rosenthal, Biesecker and Biesecker (2001) helped to inform the design of the interview form in my own work.

Rosenthal, Biesecker and Biesecker (2001) conducted a qualitative, descriptive study to understand more about the ways in which lack of diagnosis affected parental coping and adjustment to their child's condition. Mothers and fathers of children with multiple congenital anomalies suggestive of an underlying syndrome, for whom it was not possible to make a definitive diagnosis, were recruited through a National Human Genome Research Institute in the United States. While there may have been functional and descriptive terminology for individual features of the child's condition, there was no unified diagnosis or etiological explanation. Key areas where parents perceived a diagnosis would have an impact were identified as: labels, causes, prognosis, treatment, acceptance and social support (Rosenthal, Biesecker and Biesecker, 2001). Parents saw the value in having a label as a way of helping others understand their child's condition and improving access to services. Parents were also concerned about reproductive risks and assigning blame or understanding what caused their child's disability, with some parents feeling diagnosis would resolve issues of personal responsibility. The majority of parents expressed a desire for a diagnosis that would provide insight into their child's prognosis, both developmental progress and long term outlook including life expectancy. Access to treatment, acceptance of their child's condition, and access to social support, including contact with other families, was perceived to be affected by not having a diagnosis. Few parents had any contact with other parents of children with unidentified syndromes.

The Rosenthal, Biesecker and Biesecker (2001) study established that absence of diagnosis could have a significant impact on parents, affecting their response to their child's condition, and that the significance of diagnostic information may be complex. They reported that parents were highly motivated to obtain a diagnosis when their child was young and recalled an intense desire for diagnosis at this time. They also reported a strong affinity for tentative diagnosis, with subsequent difficulties when these diagnoses were later ruled out. During the interviews, parents frequently compared themselves to the parents of children with well-known conditions, such as Down syndrome or Cystic fibrosis. They contrasted their own sense of isolation with the situation of those parents of children with more familiar conditions. Describing the situation of undiagnosed disability in this context as one of the most common and unsatisfying situations encountered in medical genetics clinics, the need for further research was suggested in multiple areas.

Rosenthal, Biesecker and Biesecker's (2001) study was restricted to parents of children with 'an unexplained MCA syndrome' so included only children who were believed to have a genetic disorder. This was the first study examining the significance of diagnosis for parents of undiagnosed children. The authors did not conduct any further research in this area, although suggest that anecdotally they continued to find that diagnosis was commonly of great significance for parents (Eric Rosenthal, personal communication, 6<sup>th</sup> April 2004). The study by Rosenthal and colleagues did not include any sociological consideration of diagnosis or its absence. It was a significant precursor to my own research work with families living without a diagnosis, however I was keen to move on from this work by including relevant theoretical work to situate it in its sociological context alongside carrying out the empirical study.

Lewis, Skirton and Jones (2010), following on from this work by Rosenthal, Biesecker and Biesecker, (2001), carried out a study exploring the parental experience of raising a child without a diagnosis. They conducted interviews with 14 parents of 9 children recruited through a large regional genetics centre in the UK. They identified a gap in the literature relating to studies conducted in the UK investigating the experience of parenting a child without a diagnosis. They describe their study as an exploratory study of parents' feelings and experiences and they used a qualitative method with a grounded theory approach. The overriding theme they identified was that the experience of having an undiagnosed child



was a 'journey' encountered by parents, a journey which began with the identification of a medical problem and which was still on-going at the time of the interviews. They found that a number of experiences described by parents were common to families raising a child with a named diagnosis, and suggested that lack of diagnosis added a further layer of complexity to an already difficult situation.

Results of their research with families generally echoed those found by Rosenthal, Biesecker and Biesecker (2001). Two of the most common reasons given by parents for wanting a diagnosis were for 'help with care and treatment' and 'to know about the future'. Parents also cited wanting to have 'a reason' as a motivating factor in seeking a diagnosis. A diagnosis was also seen as a social tool by a number of parents, who had difficulty describing their child's disability to others. They used a 'social diagnosis' like development delay to manage social interactions in the absence of diagnosis. Other reasons cited for wanting a diagnosis included concern about recurrent risk in future pregnancies, to help with their child's education, and to have an explanation to give to their child. All parents said they would like a diagnosis, although many said it was no longer as much of a priority.

Lewis, Skirton and Jones (2010) highlight the emotional impact of living without a diagnosis for parents. They suggest that parents felt a lack of control, particularly with regards to their child's future, and experienced uncertainty of prognosis in both the short and long term. Some parents expressed a sense of guilt and an emotional rollercoaster was also described by a number of parents, with lows but also highs. One of the coping strategies used by parents was looking for information, either online or via other parents, and taking an active role in ensuring their child received the best care and treatment. They also report that parents wanted to be active participants in the diagnosis process and to be acknowledged as experts on their child's condition. The authors identified that the issue of frustration was a particularly prevalent aspect of the experience of living without a diagnosis.

This study, published after I had carried out my interview study, is highly relevant to my own work as one of a very small number of studies that looks at the significance of diagnosis for families with disabled children 'living without a diagnosis'. However, I found it difficult to make sense of the conceptual framework of the study. The authors identified a journey with two components: the *inner emotional experience* (the realisation that there's a problem,

the experience of testing, reasons for wanting a diagnosis, emotional impact, and active coping mechanisms) and *the outer sociological experience* or *sociological journey* (experience with professionals, accessing support networks, and issues like education and housing). I did not feel this method of organising their data analysis made sense, in part because of the misleading use of the term 'sociological'. Categorising 'emotional impact' as an inner emotional component of the experience of living without a diagnosis, and 'accessing support networks' as part of the 'sociological journey' was neither explained nor appeared logical. Despite this confusion, the results were meaningful in that themes were identified that further suggested not having a diagnosis came with its own unique impact and that further research in this area was needed.

The same authors (Lewis, Skirton and Jones, 2012) carried out a more recent study to develop an evidence based psychosocial information booklet for parents of children without a specific diagnosis. They drew upon research data from their earlier study (Lewis, Skirton and Jones, 2010) as well as a literature review of the research with families living without a diagnosis. Their literature search revealed that very little information was available for parents of children whom it is believed have a genetic condition but are without a specific diagnosis. They assert that parents of undiagnosed children are likely to have distinct information needs from parents of children who do have a diagnosis.

This work (Rosenthal, Biesecker and Biesecker, 2001; Lewis, Skirton and Jones, 2010; Lewis, Skirton and Jones, 2012) confirms that there is as yet little research looking at the parental experience of living without a diagnosis, and suggests what issues may potentially be salient for parents of undiagnosed children. However, this work omits to consider absent diagnosis beyond the empirical realm. No study to date considers absent diagnosis for childhood disability from the perspective of the sociology of diagnosis, nor how absent diagnosis in this context remains under-theorised. In taking this work forward, it was felt to be timely and important to consider both the lived experience of absent diagnosis in greater depth, and also the relevant theory relating to non-diagnosis.

As there are very few studies looking directly at the experience of living without a diagnosis in this context, and given the lack of consistency in definitions and terminology in research work looking at childhood disability, it is important to look wider than those studies dealing

exclusively with the parental experience of living without a diagnosis. Findings from other studies with families that compare the experience for families with and without a diagnosis are also relevant, as are studies where absence of diagnosis is considered among other aspects of the experience of parenting a disabled child, or where there has been significant delay in obtaining diagnosis.

Studies comparing the experience of parents with and without diagnosis suggest that uncertainty or ambiguity of diagnosis can increase emotional stress and impact on coping for parents (Graungard and Skov, 2007) and result in parents receiving little information and having difficulties accessing support (Makela et al., 2009). In one study, parents of children with a 'mental retardation of unknown etiology' showed broad psycho-emotional disadvantages compared to mothers of non-disabled children or children with a common diagnosis (Lenhard et al., 2005). Graungaard and Skov (2007) found the emotional reaction of parents to be highly influenced by the diagnostic process and that without diagnostic certainty parents had difficulties coping with an uncertain future. The parents' use of coping strategies was found in their study to be strongly related to diagnostic certainty, and parents' experience and coping possibilities when realising their newborn child was disabled was found to be strongly influenced by the nature of the diagnostic process and by the certainty of diagnosis. Other studies that consider absent diagnosis highlight that parents who experience greater uncertainty feel less control over their child's medical condition which may result in less effective coping and poorer adaptation (Madeo et al., 2012).

Concern about the future and their child's prognosis can be significant for parents of children with uncertain diagnosis. The quest for a diagnosis can be a quest for knowing what the future might bring; a quest for prognosis (Graungaard and Skov, 2007), a need for information that could help parents plan for their child's future (Makela et al., 2009) and gain knowledge about future expectations and appropriate interventions (Watson, 2008). Parents of disabled children with uncertain diagnosis may have concerns about the future stability of their child's condition; their child's future limitations; short and long term planning; recurrence risks; the care of the child when they are no longer around; and isolation in the absence of anything to base expectations on (Madeo et al., 2012). Uncertain diagnosis can impact on parents' ability to look forward to prognosis, and also to look back to aetiology. Knowing and understanding the cause of their child's disability is important for

parents (Watson, 2008). Parents may desire an aetiological diagnosis so that they can assess the risk of recurrence (Graungaard and Skov, 2007). The desire to obtain a causal diagnosis has been reported to be at its most intense when symptoms first emerge (Makela et al., 2009). Lenhard et al., (2005) report that diagnostic and prognostic uncertainty is one of the major psychosocial stressors for parents of children with disabilities or other acute and chronic conditions and that diagnosis can assist parents in making reproductive choices (Lenhard et al., 2005). Diagnosis may also resolve parental feelings of guilt or blame for their child's condition (Lenhard et al., 2005; Makela et al., 2009; Watson, 2008).

Diagnosis may also be perceived by parents as enabling access to services or therapies at school (Makela et al., 2009; Genetic Alliance UK, 2010) and access to social support in that parents may have difficulties finding other parents to share their experiences with (Genetic Alliance UK, 2010). Having a name for their child's condition can assist parents in accessing formal and informal support and it may be easier for parents of children with a specific diagnosis to join or form associations, than mothers of children with a vague or completely uncertain diagnosis (Lenhard et al., 2005) and to access specific services (Watson, 2008). Parents of a child without a diagnosis may perceive disproportionate amount of services available to children with common conditions like 'autism' (Makela et al., 2009) and not having a diagnosis can impact on establishing emotional companionship with other families (Makela et al., 2009) and obtaining contact and support from others who have been on the same journey (Madeo et al., 2012 p1882). For parents of children with undiagnosed medical conditions (including chronic illnesses and disability) isolation can be experienced with reference to knowing what to tell others, inability to gain the acceptance of others, and having a lack of models on which to gain expectations (Madeo et al., 2012).

While the core literature identified above is seen to be relevant to the study of living without a diagnosis in that it examines the experience of absent diagnosis, other research work with families who experience a long delay in obtaining a diagnosis can also offer insights. The difficulty of obtaining a diagnosis of autism is highlighted by Midence and O'Neill (1999). For children with Autism Spectrum Disorders (ASD) the path from a parent's early concerns to a diagnosis can takes several years (Lord and Luyster, 2006). Research has consistently found a significant lag in time from the onset of ASD symptoms and when an autism diagnosis is made (Miller, Pandey and Berry, 2014; Sivberg, 2003; Howlin and Asgharian,

1999; Wiggins, Baio and Rice, 2006). This 'lag time' (Chan et al., 1994) between symptoms onset and diagnosis can lead to frustration and have consequences for the child and family (Howlin and Asgharian, 1999) and some parents potentially even considering the 'lagtime' years, the years spent waiting for diagnosis, to have been wasted (Carmicheal et al., 1999).

Parents may experience self-blame and guilt in the absence of an explanation for their child's difficulties (Midence and O'Neill, 1999) and mothers may view diagnosis as a vindication of blame (Williams, 2007; Carmicheal et al., 1999) and a validation of their child's difficulties (Avdi, Griffin, and Brough, 2000). Diagnosis can also be seen as a way of accessing help for their child (Williams, 2007) and improving access to support and assistance (Midence and O'Neill, 1999). Financial help, appropriate intervention, contact and support from other families, and information provision are among the benefits perceived to be associated with diagnosis for parents experiencing diagnostic delay for their child (Carmicheal et al., 1999). Parental isolation and coping may be improved with diagnosis (Williams, 2007; Midence and O'Neil, 1999) and difficulties in public spaces with describing their child's difficulties to others may be resolved (Midence and O'Neill, 1999; Avdi et al., 2000). Knowledge about prognosis, causes and treatment may be perceived by parents as being more available with diagnosis (Avdi et al., 2000).

Studies looking at delay in diagnosis, such as these, offer insights into how diagnosis is perceived by families. While families in this research may go on to receive a specific diagnosis for their child, their attitude to diagnosis in the lag time period prior to diagnosis is relevant to the study of living without a diagnosis. Every illness goes through a 'diagnosis deferred' stage (Lossos et al., 1989) in which patients are passing through the diagnostic funnel and medical assessment is working up to diagnosis. Prior to classifying patients into a specific nosological grouping, they exist in a liminal state of lag time. Certain conditions may be more difficult to diagnose, particularly in young children, and so families may experience a period of sustained lag time. For parents of children with very difficult conditions to diagnose, like the parents in my own study, this lag time may be infinite.

## 2.5 Conclusion

In this chapter I have reviewed a wide range of both theoretical and empirical literature concerned with diagnosis; including literature relevant to the sociological study of diagnosis, studies of the process of paediatric genetic diagnosis, and empirical research work with families. This literature will be synthesised with my own research work throughout this thesis. As described by Boell and Cecez-Kecmanovic (2010, 2014), the literature review was an iterative process; evolving over time. The reading of the literature, conducting of the research, and writing the thesis was not a linear process, but was on-going. Some of the literature introduced in this chapter was gathered prior to the interview study, for example the research with families that was carried out prior to 2006 and the work deconstructing medical classification systems. The literature review also developed beyond the time of the interview study, however, and so this review represents a journey over time. Two areas in the literature, in particular, were brought in as a result of the iterative process of returning again and again to the literature to mine new knowledge: the work on dysmorphology and the process of paediatric genetic diagnosis, and the work since 2009 on the sociology of diagnosis. Also in qualitative work with families with disabled children, there was a small growth in the work directly related to absent diagnosis in the time period after the interview study. My understanding about diagnosis and its absence has evolved over time, on this hermeneutic journey of learning. In the next chapter, still holding the theme of journey, I will describe the methodologies informing the research and the methods of the study design and the data analysis.



## **Chapter 3      Methodology and Research Methods**

### **3.1      Introduction**

### **3.2      Methodologies informing the research**

*The Methodological Journey*

### **3.3      Ethical Considerations**

### **3.4      Research Design and Methods**

*Recruitment of participants*

*The study design*

### **3.5      Methods of data analysis**

*Producing transcripts*

*Developing themes*

### **3.6      Personal Reflections**

### **3.7      Conclusion**



### 3.1 Introduction

*I interview because I am interested in other people's stories*

(Watkins, 1995, p24)

The motifs of story and journey permeate this thesis. In this chapter, I tell the story of the methodological journey of the study. I then consider the research design and methods. I took both a narrative and thematic approach to data collection and analysis. Chapter 5 describes the narrative approach and analysis; in this chapter I focus on thematic analysis, the principal method of analysis. The study was a qualitative interview study. The interview, and the narratives told therein, is held to be at the crux of inquiry into making sense of human experience.

Interviewing is a basic mode of inquiry. Recounting narratives of experience has been the major way throughout recorded history that humans have made sense of their experience (Seidman, 2006, p8)

Interviews were in-depth and semi-structured. The study sought to understand the lived experience (Shutz, 1967; Van Manen, 1990) of participants and the meaning they made of that experience. In this chapter I first describe the methodologies informing data collection by recounting my methodological journey (3.2). I then consider the ethical considerations of the study (3.3), including details of ethical approval granted by the appropriate Research Ethics Committees. In considering the research design and methods in the next section (3.4), I describe the recruitment of participants and the study design. I then explain the methods of data analysis (3.5) looking at how transcripts were produced and how themes were developed. I offer further personal reflections on the research in 3.6, before concluding the chapter.

### 3.2 Methodologies informing the research

Here, I describe the journey of deciding on the right methodologies for data collection. When I started out at the beginning of this research journey, I was in search of the *right* methodology; trying to find a singular, stable approach that could be used as the best method of researching with parents and of finding out about the experience of absent diagnosis. In the end, I learned that the methodologies I needed were multiple, changing and hybrid. Rather than finding a single methodology that was uniquely right for informing my research, instead I learned that aspects of multiple methodologies were relevant; and that in being fused and overlapping they formed a composite that became my methodological approach. This resulting methodological bricolage (Denzin and Lincoln, 2000) informed the design of my research. Denzin and Lincoln (2000) suggest that the researcher's role may be one of methodological, theoretical, interpretive, or narrative bricoleur, with their finished product of research a quilt-like bricolage.

The product of the interpretive bricoleur's labour is a complex, quilt-like bricolage, a reflexive collage or montage; a set of fluid, interconnected images and representations. This interpretive structure is like a quilt, a performance text, or a sequence of representations connecting the parts to the whole (Denzin and Lincoln, 2000, p6).

Etherington (2004) refers to this bricolage, in the context of the exploration of different methodologies, as often underpinned by a heuristic journey of discovery. I saw the process of exploring methodologies as a journey of learning. I have therefore chosen to describe this methodological journey and my self-reflection during this journey, rather than merely offering definitions of the methodological perspectives that, in the end, informed the data collection.

Steiner Kvale (1996) uses a journey and story metaphor to describe the qualitative interview researcher, depicting the researcher as:

A traveller on a journey that leads to a tale to be told upon returning home...The interviewer wanders along with the local inhabitants, asks questions that lead the subjects to tell their own stories of their lived world, and converses with them in the original Latin meaning of conversation as 'wandering together with' (Kvale, 1996, p4)

His words are evocative of a quest narrative, with the researcher returning home with a story to tell. They are also reminiscent of the vernacular poetry genre telling of sharing narratives, an ancient example being Chaucer's *Canterbury Tales* (late 14<sup>th</sup> Century) in which a story is told about stories as the poet travels alongside the pilgrims.

In my own journey, seeking stories and telling a story about stories, I started with the belief that a single methodology was possible with which to best approach and know the data. At the beginning of my research journey, therefore, I embarked on a thorough search of the methods literature seeking this definitive methodology; anticipating the right approach. I consulted key social research authors (Denzin and Lincoln, 2000; Holstein and Gubrium, 1995, 1997; Gubrium and Holstein, 2001, 2003) who uphold the interview as (inter)active, and interview participants as meaning makers rather than passive vehicles for retrieving information from an existing vessel of answers (Holstein and Gubrium, 1995). I was drawn to critiques of the detached interviewer and the myth of scientific interviewing that suggested that the interview could be seen as an interactional event based on mutual knowledge (Cicourel, 1964). As the mother of a disabled child without a diagnosis, the subject of my research was personally relevant, and I embraced the notion of the interview as collaborative.

Silverman (1993) describes our 'interview society' (1993, p19), highlighting the ubiquity and significance of the interview format in our lives and that the interview is central to making sense of our lives. The interview is held to be a site of meaning production (Gubrium and Holstein, 2001), and the characteristic format for personal narrative (Atkinson and

Silverman, 1997). Qualitative interviewing is described as a kind of guided conversation (Kvale, 1996; Rubin and Rubin, 1995) with researchers asking the questions and listening for the meaning of what is being shared. Kvale defines qualitative research interviews as 'attempts to understand the world from the subjects' point of view, to unfold the meaning of peoples' experiences, to uncover their lived world prior to scientific explanations' (1996, p1). Given that I was seeking to understand the experience of living without a diagnosis from the participants' point of view and the meaning of that experience, a qualitative approach was needed and an interview study was most appropriate for gathering in-depth data.

I began to consider what kind of qualitative interview study would be most relevant. I was keen for my interviews to be 'active' (Holstein and Gubrium, 1995) and to avoid the familiar asymmetrical interview relationship of the traditional research model (Gubrium and Holstein, 2001). I believed that, as Gubrium and Holstein describe, the active interview would yield rich data, a suggestion eloquently phrased by Terkel.

There were questions of course. But they were casual in nature. In short, it was a conversation. In time, the sluice gates of damned-up hurts and dreams were open. (Terkel, *Working*, 1972, cited in Gubrium and Holstein, 2003, p30)

I wasn't convinced, however, by all aspects of the active interview process Holstein and Gubrium (1995, 1997) describe, and felt concerned about the 'guiding' approach of the interviewer and the way the interviewer was encouraged to provoke and direct responses from participants. I was planning in the initial stages of the interviews to ask my participants to tell their own story, and I didn't want to do this by imposing frameworks on how they told their story. So while I took on board many of the aspects of active interviewing, I still sought a methodology more apposite for the kind of research I intended. I kept open the idea of active interviewing, but without these elements.

I learned that I wanted my interviews to be in-depth. Van Manen (1990) proposes that the purpose of in-depth interviewing is not to test an hypothesis, but to understand the lived experience of other people and the meaning they make of that experience. I sought to interview in a way that not only enabled depth, but also allowed the nuance and complexity of data to be captured (Mason, 2002) and that would be generative in that new knowledge would be produced (Legard, Keegan and Ward, 2003) as well as a priori themes explored. I

wanted to find out about the lived worlds of the participants, in Kvale's words I wanted the interview to help me 'understand the world from the subject's point of view, to unfold the meaning of people's experiences, to uncover their lived world' (1996, p1). Kvale calls the interview an 'Inter View' in that what takes place is 'an inter change of views between two persons conversing about a theme of mutual interest' (Kvale, 1996, p2). This aspect of mutuality is also reflected in the work of other research methods authors, for example Mishler (1986) who regards the interview as an interactional accomplishment and Johnson (2001) who suggests the interview is a conversational partnership.

In-depth interviewing demands a greater involvement of the interviewer's self than other forms of interviewing. Mutual self-disclosure and trust is progressively built; this requires that the interviewer offers some form of reciprocity, for example sharing their own views, feelings, or reflections on the interview topic (Johnson, 2001). In-depth interviewing seeks deep information and understanding, aiming to explore the contextual boundaries of the experience or perception being studied, to uncover what is normally hidden from ordinary view to penetrate more reflexive understandings about the experience being studied (Gubrium and Holstein, 2001). Although I welcomed the prospect of mutual disclosure and sharing mutual stocks of knowledge, and of reflexively gaining deep understandings of the experience of absent diagnosis for parents, I didn't wish to stick rigidly to the suggested study design for in-depth interviewing. For example, it is recommended to ask 8 or so open questions and to finish the interview by summing up responses for participants. As I had a priori themes to explore, as well as anticipating that new themes may be generated, I felt restricting myself to a small number of very open-ended questions would not give me the breadth of understanding I sought. However, I felt committed to carrying out interviews that were face-to-face, flexible in structure, interactive, generative and to using probes to seek depth of answers (Legard et al., 2003).

I also explored the grounded theory approach, through the work of Glaser and Strauss (1967), and Strauss and Corbin (1998), and considered the development of codes from the data rather than from theory and a priori assumptions. However, having already conducted interviews with parents living without a diagnosis as part of my MSc dissertation, and having reviewed other relevant literature, I realised that I had a priori ideas about what kind of things I'd like to *know* about the experience of not having a diagnosis. While I wanted to

give the parents the space to tell me about their experiences in their own words, and for new themes to be generated from the data beyond these a priori themes, there were also several areas of research interest I wanted to ask about. I was reluctant to allow only concepts emergent from the data to dictate the direction and the nature of the data collection. While I didn't wish to approach the data collection with a preconceived and verificational approach, compelling it to yield the results I desired (as Glaser suggests 'if you torture the data enough, it will give up', Glaser, 1992, p123), I did wish to *know* about aspects of the parents' experiences I was interested in, as well as hearing their stories. I concluded that a mixed approach of both guiding conversation around previously identified key themes, as well as allowing themes to emerge spontaneously in the data, was more desirable for the study.

I embraced the idea of starting where you are (Lofland and Lofland, 1995). Lofland and Lofland (1995) advocate this idea of starting where you are, by which they mean researchers should consider studying the social phenomena they have ready or advantaged access to. I identified with the idea that I was carrying out complete-member research (Adler and Adler, 1987) in which the researcher is usually fully committed to and immersed in the groups they are investigating. Further to this, I also identified with Adler and Adler's (1996) description of the parent-as-researcher role, where the parent has dual membership, with benefits of this dual role (and indeed ethical considerations) similar to complete-member research. This description, however, still did not define all aspects of my desired research methodology and so I continued on my search, having learned that I may need multiple methodologies.

In further considering the relationship between the researcher and research participants, I examined feminist approaches to interviewing. Feminist methodologists have questioned the traditional scientific method as being appropriate for capturing human experience (Harding, 1987) and place emphasis on power relations, with early feminist researchers suggesting the power relation between the researcher and the researcher should be non-hierarchical (Skeggs, 1994; Maynard and Purvis, 1994). Feminists have rejected the inevitability of the power hierarchy between researcher and participants, instead arguing for the significance of a genuine rapport (Maynard and Purvis, 1994). Feminist researchers may reject the conventional stance of the interviewer trying to maintain a distance from the participant by not disclosing their experiences, feelings or opinions to the research participant. Instead interviewer self-disclosure; when the interviewer shares ideas, attitudes

and experiences with participants, may be encouraged with many feminist researchers choosing research topics that are deeply personal. This can make the interview process emotionally intense for both the interviewer and the participant, and can lead to self-disclosure.

In practice, it may be difficult to know what kind of disclosure is appropriate in the research interview (Reinharz and Chase, 2003). Reinharz and Chase advise to consider carefully whether, when and how much disclosure makes sense in the context of particular research projects and with specific participants, asking: When does self-disclosure indicate the sharing of power within the research relationship, and when does it indicate that the researcher prefers to speak rather than to listen? (2003, p80). Janet Parr (Parr, 1988), referring to her interviews with mature students, describes how having similar personal experience to the participants can be both enabling and limiting with reference to participants' voices. She says, 'my natural inclination was to be open and honest...about my own position, but I wanted what they told me to be their own story, not a reflection of my own' (Parr, 1998, p91). Empathy is described as a 'double-edged sword, both enabling and disabling' (1998, p99).

While the possibility of self-disclosure in the interview setting had resonance for the kind of methodological approach I intended to take, I kept these cautions in mind. As well as choosing a personally relevant research topic, and being open to self-disclosure in the interview setting, many of the other characteristics associated with the methods, practices and epistemologies of what was identified as feminist social research were relevant to the kind of research I wanted to carry out; the rejecting of power hierarchies between researcher and researched, a concern to record the subjective experience of research, concern with reflexivity, and a focus on ethical questions. I wanted to incorporate these qualities into my own research work with parents. Therefore, I assumed many of these characteristics as forming part of the methodology for my own research.

Reflexivity has become an increasingly compelling theme in contemporary social research (Etherington, 2004) and a research topic often has personal significance for the researcher. Warren (2001) describes the interview, like ethnography, as about self as well as other and the purpose of qualitative interviewing and associated fieldwork is to understand others'

meaning making and also how those meanings intersect with our own story. This thesis is about the researcher's self as well as others; and in understanding the meaning making of the parents, I also consider my own story.

Having met with the concept of the 'active interview' (Holstein and Gubrium, 1995, 1997) further on in my methodological journey I encountered the 'interactive interview' (Ellis, Kiesinger and Tillman-Healy, 1997). Ellis et al. (1997) describe interactive interviewing as a method of obtaining an in-depth and profound understanding of people's experiences with sensitive and emotionally charged subjects such as loss, childbirth, and illness. They view interviewing as a collaborative communication process, with both participants and researcher sharing personal and social experiences. This, they contend, allows the distinction between researcher and participants to be blurred, with the researcher's disclosures encouraging the participants to be more open themselves in the interview setting. They say: 'The feelings, insights, and stories that researchers bring to the interactive encounter are as important as those of the respondents' (1997, p121). Interactive interviews are described as lengthy, often multiple, with a focus on building a relationship of mutual self-disclosure and trust, and on the emotional and the intimate nature of the encounter. Researcher involvement is seen to promote dialogue, rather than interrogation (Bristow and Epser, 1988). Reflexivity regarding the research process is part of the methodology, and personal experience of the topic under investigation is advised.

Prior to conducting my research, I was eager to adopt an interactive approach, although I was unsure about the level of disclosure expected and whether this would enhance or detract from participants' own stories. I was pleased, therefore, to encounter the work of Ellis and Berger (2003) who describe various types of collaborative interview, including 'reflexive dyadic interviewing'. In this methodology I found a balance between the full disclosure practices of interactive interviewing, as described by Ellis, Kiesinger and Tillman-Healy (1997), and the traditional model of more rigid separation of researcher and respondent with no disclosure on the part of the researcher.

Reflexive dyadic interviewing, as described by Ellis and Berger (2003), follows the more typical protocol of the interviewer asking the questions and the interviewee answering them, but the interviewer will usually share personal experience about the research topic or may



reflect in the interview on the process of the research. The interviewer may often feel a reciprocal desire to disclose, for example when telling the participants about the research the interviewer may reflect on the personal reasons for choosing the research topic and their emotional response to the research. Rather than seen as a hierarchical question and answer exchange, the interview is held to be more of a conversation than an interrogation and interview style is conversational. Ellis and Berger describe other forms of collaborative interviewing, including interactive interviewing and co-constructed narratives, forms that include the researcher as a central character in the interview story (Ellis and Berger, 2003). The middle ground offered by the reflexive dyadic approach, in which the researcher may share personal experience with the participant while the interview retains the question-answer structure, appealed. I felt I had found a methodology highly suited to my anticipated research study, and the design of my research was informed by my chosen methodologies. These chosen methodologies, or aspects of them, formed a methodological bricolage; qualitative, semi-structured, in-depth, active, reflexive dyadic, narrative, thematic, collaborative, generative, ethical, empathetic, parent-as-researcher, complete member research, interactive, an Inter View. With this montage of methodologies, I proceeded to design and carry out my interview study.

One of the vital components of the collaborative research methodology I had intended to use was self-disclosure. While the reflexive dyadic approach described above laid less stress on complete collaboration of the interviewer, it still anticipated significant self-disclosure. However, even over the course of the first few interviews it became apparent that my self-disclosure seemed to interrupt the flow of the parents' own story, and I intuitively restricted my self-disclosure. I responded to questions asked by the participants about my own experience but did not talk in-depth about my experiences. That the participants knew I was the mother of a disabled child, and further a child who did not have a diagnosis, seemed beneficial to the interview process. Parents talked about therapies, equipment, disability related welfare benefits, medical conditions, tests and assessments and named consultants and medical centres I was familiar with. Anything I wasn't familiar with, I asked about, so the participants were aware that I had some mutual shared knowledge, but not exactly the same knowledge for our journeys had been different.

I suggest that our mutual stocks of knowledge were beneficial to the process of data collection; parents described their lives with their child in a detailed, open way and I naturally had empathy with the parent and a good relationship seemed to develop between us. However, self-disclosure was much more limited than I had expected. The mutual experience and knowledge we shared remained in the background, sometimes coming to the foreground briefly, for example if I made a comment about a particular treatment, test, service, medical condition or piece of equipment in response to their narrative. For example if a parent talked about their child wearing leg splints, I might share that my daughter also wore splints; or if they talked about a 'feeding tube' I might say that my daughter had a gastrostomy until she was two years old. On these occasions, when I was not directly asked about my own experience, I gave only a little background information so that the parent knew of this mutual knowledge. My feeling was that this facilitated the research process in that it had time saving benefits. Instead of having to define and explain different equipment (like walkers or standing frames), services (like direct payments or respite care), treatments (like orthotics or oxygen therapy), or medical conditions (like hypotonia, hydrocephalus, apnoea), the shared knowledge potentially meant the parent could spend more time talking about their experience and their response to it in an in-depth way. However, beyond the mutual knowledge in the background and the limited moments of self-disclosure, there was not the interactive element in relation to researcher self-disclosure that had been anticipated when preparing for the interview study. Parents rarely asked me to share details of my own life. They were keen to share their own stories and parents often talked extensively about their experiences. I was, after all, there to hear their story. If I was to learn about their lives in the level of detail I wanted to, there was no place in the interview encounter for the telling of my own story. Therefore, in the interviews I only shared what I felt was necessary to improve the research relationship and to allow the parent to talk on a level of mutual understanding with me.

The disclosing of information had to be thought through carefully. Being able to empathise with the parents, by recounting my own similar experiences, was – as Parr (1998) described – a double edged sword, and I had to be careful to share enough information that dialogue was enhanced, but not so much that the flow of the parents' own story was interrupted. As Frank (2002) cautions, when you're part of the research community you're studying, there needs to be a balance being knowing too little, and knowing too much. In the end then,

while I was reflexive about the interview process, the interviews were less interactive than I had anticipated. The data was enhanced by my complete-member status, but I didn't indulge in excessive self-disclosure. I heard the parents' stories: they were told, damned-up hurts and dreams and other things besides. The interview guide I had prepared and the informal yet semi-structured setting of the interviews generated rich data about the parents' lived experience. Interviews were friendly and a good rapport was established, without the apparent need for extensive self-disclosure of the researcher.

The contemporary social researcher can scaffold their research with one of the many labels that describes how we do research. There is a proliferation of categories of methods and methodologies in social science that describe a plethora of approaches: some are labels for general approaches to social research, some are specific strategies for research design and analysis. Atkinson warns there is a danger of turning 'the pedagogical half-truths of text book knowledge into prescriptions for research practice' (Atkinson, 1995, p123). I began my research armed with such textbook knowledge, and learned during the process of the research that sometimes the methodology is born from, or at least evolves with, the practice of actually doing the research. According to Kvale (1996), the design of qualitative interview research is open-ended in the sense that it is more concerned with being attuned to who the researcher is travelling with, than with setting out a precise route for all to follow.

I have shared a story of my journey to find the right methodology for the study. In the end, I didn't find a precise label for the methodology I used, a label that would suggest all the different aspects of the approach. Despite not having a definitive label to lean on when describing the methodological approach, I am confident that my methodological journey was an indispensable one for the research and that the resultant use of a fusion of modified methodologies, my methodological bricolage, yielded data that was generous, profuse and exciting. In this next section of the chapter, I describe the ethical considerations of the study.

### **3.3 Ethical Considerations**

The ethical issues related to the study were thoroughly considered and multi-site ethical approval was applied for and granted by the relevant Research Ethics Committees. Relevant documentation submitted to the Research Ethics Committees and used in the research is included in the appendices; the consent form (Appendix 3) and the interview guide (Appendix 4).

This study required very careful consideration of ethical issues. One of the main concerns was with anonymity. I was aware that, in the interviews, parents would be likely to describe their child's difficulties and a set of medical symptoms or clinical characteristics that were unique to their child; so unique that they did not fit the pattern of any known diagnosis. Merely changing names and other personal identifiers was not going to guarantee anonymity. While anonymity is an issue that needs consideration in all qualitative interview studies, it was even more pertinent in this research that included participants by virtue of the exclusiveness of their child's condition. Such exclusivity could potentially make them more easily identifiable. To address this issue I made every effort to modify personal identifiers to reduce the possibility of identification. In addition to using pseudonyms, for example, I also changed the names of very rare syndromes the child was reported as being tested for as well as other details that might enable a reader to identify the family. I also felt it was important to explain to the parent that, while every effort would be made to ensure anonymity, anonymity could not be guaranteed. Parents were asked to sign a consent form (included in Appendix 1) that stated clearly that anonymity could not be guaranteed. In going through the consent form with the parents, I drew attention to this point about anonymity. This enabled parents to decide what they wished to say and share with me in the interview, fully informed that their anonymity could not be guaranteed.

The potential for emotional harm for participants was another issue that required careful consideration. As with the issue of anonymity, this is a common ethical consideration when conducting qualitative research, particularly if interviewing on sensitive topics. Many of the parent participants had experienced anxiety, distress, and sorrow and recounted experiences of difficult times including serious illness, hospital stays and difficulties associated with caring. Recounting such experience can provoke powerful emotional responses. If a parent

was upset during an interview, I offered supportive words and empathy. I also reminded the parent that they could stop the interview at any time, and made sure they were happy to continue with the interview before moving on. Upset caused by describing their experiences or their child's difficulties was seen as a normal human response to difficult experiences. As a parent myself, who has experienced long stays in hospital and the chronic illness of my daughter, I recognised the value of empathy in this situation. I had a pre-prepared list of support organisation ready to share with the parent should they seem to require it. Parents gave positive feedback about their experience of taking part in the interview and most commented positively on the interview experience without prompting. Parents were offered a copy of the transcript of their interview, which around a quarter opted for.

An additional ethical issue that arose during interviewing was the presence of the participant's child during the interview. On four occasions the child was present during the interview. Although I did make every effort to ensure I could interview the parent when the child wasn't there, sometimes this wasn't possible. Prior to starting the interview I discussed the presence of the child with the parent, agreeing language use (whether to avoid certain words) and getting an idea of the level of understanding of the child. On all occasions the parent reassured me that their child would not understand what was being said or would not be upset by what we were talking about. I decided that the parent was best placed to decide whether or not their child should be present during the interview, and accepted their responsibility as paramount when it came to deciding whether the child was at risk of being upset by overhearing our interview. Most of the time, the child played in a nearby room. None of the children became distressed during the interview process and all seemed to enjoy my presence.

Gaining ethical approval was a key part of the preparation for the research. This was seen as a time consuming but necessary aspect of beginning the data collection for the study. Approval was sought from the relevant NHS Research Ethics Committees (RECs). As the intention was to recruit parents of children who attended clinics in different geographical areas, it was necessary to apply for multi-site ethical approval. The application for approval was successful with only a few minor amendments. The study was approved by both relevant local research ethics committees with no difficulty and in good time. When I sought approval from the relevant Research and Development Offices, however, I encountered

some difficulties. It took a very long time to progress the application for a number of reasons, including difficulty obtaining reliable contact with consultants willing to act as a contact within the NHS site. By the time the Research and Development office had finalised the approval for the study at their site, more parents than needed had been recruited for the study via the more informal advertising in a monthly magazine for parents of children with additional needs. I awaited full approval from all relevant parties prior to beginning the research with parents, although did not need to recruit parents via the consultant contacts as I had originally anticipated.

Overall, the experience of applying for and obtaining NHS ethical approval was a very positive one, with the relevant Research Ethics Committees readily granting approval. Had it been necessary to recruit parents via consultant contacts, however, the interview study would have been significantly delayed. The process of applying for ethical approval was very useful in that it encouraged me to carefully think through and plan all interview guides, contact letters with parents, consent forms and advertising for the study. With the relevant ethical approval in place, the interview study could proceed. In the following section, I look at the research design and methods for the study.

### **3.4 Research Design Methods**

In this section I will describe the research design and methods of data collection. I will explain the recruitment of participants and the study design. As described in section 3.3, the ethical issues of the study were thoroughly considered and approval for the study was granted from the appropriate RECs. The method of recruitment of participants was different from that intended when applying for ethical approval. It had originally been the intention to recruit via consultant caseloads, however a considerable number of parents responded to advertising about the research, and recruitment via consultant caseloads was no longer necessary. The study did not begin until ethical approval was granted, and the documents submitted as part of the ethical approval application were used in carrying out the study. A table with further information about all parent participants is included in appendix 1. In this table, parents are identified by the pseudonym of the child and the child's age, educational

setting, and disability as described by the parents are detailed. Where the family lived (i.e. Central Scotland or other area/country), who was interviewed (i.e. mother, father or both) and the method, number and duration of interviews, is also detailed.

### *Recruitment of participants*

The interview study involved 26 parents; 19 mothers and 7 fathers, of children with significant and multiple disabilities for which there was no definitive diagnosis. Parents were recruited by a number of methods. The primary method of recruitment was advertising in a newsletter for families with disabled children living in Central Scotland. Nineteen of the parents were recruited in this way; via adverts in the 'Snippets' magazine produced by the Special Needs Information Point in Edinburgh and distributed to over 1500 families across Central Scotland. Following adverts in 2 consecutive editions early in 2005, 24 parents voluntarily made contact.

From the parents who made contact and were interested in being involved in the research, 19 became participants (13 mothers and 6 fathers). The remaining parents were unable to take part for a variety of reasons including: the parent changing their mind about taking part, being unable to get in touch with the parent again after initial contact, and the parent not being there when I arrived for the interview (2 parents). First interviews were carried out with parents between July 2005 and March 2006, with the majority of first interviews conducted in February and March 2006. Second interviews where appropriate followed around 6 months later (January 2006 to December 2006). All interviews were conducted in the parents' own home. Fathers and mothers were interviewed separately. It soon became clear that the interviews were yielding in-depth, rich data and interviews were often lengthy. The initial intention was to carry out two interviews with each parent; this aim was adjusted during the data collection period. Second interviews were carried out with 12 parents. Second interviews were not carried out with the remaining parents for a number of reasons. One family had moved away following the first interview; I had sent their transcripts to them and they were happy with the transcripts and interviews had yielded in-depth data about their experience and their perceptions and feelings about absent diagnosis. Three of the parents were not interviewed a second time because their experience was

atypical to the experience of the other parents in that they were not seeking a diagnosis (Rupert's parents) or there was not agreement from their child's specialist that their difficulties were medical in origin (Tom's mother). These parents, who had *atypical* experience to the rest of the parents, were interviewed once and the data collected was used in the analysis where relevant as it was felt there were aspects of their experience that emerged as significant for understanding the phenomenon under study. One of the fathers (Cameron's father) was only interviewed once as he wasn't available for interview when his wife was first interviewed, and second interviews were not carried out with Ismail's mother or Andrew and Lee's mother. Both first interviews with these parents were in-depth and lasted in excess of 1 hour (70 minutes and 90 minutes). As Ismail's mother was not his birth mother, and as Andrew and Lee were in adulthood at the time of the interview study, their experience was a little different to most of the other parents, who represented more of a homogenous group in that they were all birth parents of children rather than young adults. Kamil's mother was also only interviewed once, as our first interview was 5 hours long and there was a sense of it coming to a natural conclusion.

By the end of the first set of face-to-face interviews with parents I had 30 hours of recorded data across 19 interviews. I was beginning to notice consistency in themes emerging in interviews. I also had other parents interested in taking part in the research. I made the decision to include more parents in the research and to carry out second interviews with existing participants only if it was felt it would be beneficial. I decided second interviews would be important for those parents for whom time had ran out in the interview rather than the interview coming to a natural close. I interviewed all parents a second time who were keen to take part in a second interview and who I felt would have wanted more time in their first interview to talk about their experiences. Commonly, second interviews involved me recalling what the parent had said about diagnosis in the first interview, and probing further where relevant. Attitude to diagnosis and themes identified were widely consistent between first and second interviews, further augmenting my feeling that second interviews were not necessary for all parent participants. I include an example of a second interview guide in appendix 5.

During the main interview study described above, additional parents were making contact with me keen to tell me their story and be included in the research. These parents had



learned about the research via the support organisation Syndrome Without a Name (SWAN) USA, which had included details about the study in their newsletter after the co-ordinator of SWAN USA and I had been in touch with each other online. My initial interest in including these additional parents in the research was for the purposes of triangulation of results of the main study in central Scotland. I had been keenly following the SWAN (UK) online forum for parents for over a year and I was seeing recurring themes in parents' perceptions of the impact of absent diagnosis. I considered that I could have more confidence about the results of the main interview study by exploring whether similar themes emerged in the accounts of parents recruited via a different method. Therefore, a further 7 parents were recruited for the research in this secondary interview study (3 from Australia, 1 from England, and 3 from the USA). Methods of data collection for both participant groups are described in the following section.

The size of the sample was determined not only by the number of participants who volunteered for the research, but also by the amount of data generated in the interview study and regularity of the recurrent themes emerging as interviewing progressed. Ritchie and Lewis (2003) suggest that qualitative studies are usually much smaller in sample size than quantitative studies, as there is a 'point of diminishing return' to a qualitative sample (2003, p83). That is, as the study goes on more data doesn't necessarily lead to more information. In qualitative data, even one occurrence of a theme or code is enough to make its inclusion in the analysis framework valid (Ritchie and Lewis, 2003). Although I believed that new insights would arise with any new parent story, given they are all unique, I had a felt sense of adequate saturation of the main themes in interview data even after the first set of interviews. This is reflected in the data analysis as high numbers of participants describe the key themes (as reported in later chapters). Glaser and Strauss' (1967) concept of saturation, the point at which new data collected does not shed any further light on the topic under investigation, is referred to by social researchers as determining sample size. However, these researchers are commonly adopting a grounded theory approach to the data. While I was very aware of the recurrent nature of themes across many interviews, I did not believe a point of saturation as such had been reached as each parent tells a unique story and this will offer different insights, albeit often expressing common key themes. The final choice over the number of participants was decided by practical issues and time constraints on continuing to interview, having a large body of apparently rich data, and observing a

recurrence of themes. The total amount of recorded data was 52 hours. This data was focused on the topic of absent diagnosis, given that each parent told their 'diagnosis story' in the interview and responded to general as well as specific probes about the experience of living without a diagnosis. As a member-researcher (Adler and Adler, 1996) the flow of conversation was not interrupted by the participant having to explain aspects of their lives I was already familiar with like therapies, treatments, symptoms and equipment. The total number of parent participants was 26, with an additional interview carried out with the grandparent of an undiagnosed child and founder of SWAN in the UK (see appendix 7).

The scholarly work considering sample size in qualitative research acknowledges that qualitative work is more about cumulating an in-depth understanding of a phenomenon and its meaning than making generalisations to a larger population (Dworkin, 2012). The aim of in-depth data is to create categories from the data and examine these categories from the perspective of the 'lived experience' of research participants (Charmaz, 1990, p1162). On the issue of sample size, key social research authors and academics are often unwilling to quantify what may be a desirable sample size or at least will defend the variability of sample sizes and insist the desired number of participants depends on the study (see Baker and Edwards, 2012 for a fuller discussion). There is little definitive and unambiguous guidance about how large a qualitative research sample should be (Bryman, 2012). In the paper edited by Baker and Edwards that asks 'expert voices' to respond to the question of how many qualitative interviews is enough, there are a range of responses.

Adler and Adler (2012) state the need for a lower sample size in qualitative work compared to quantitative work. When advising graduate students they often suggest aiming for around 30 interviews, which is described as a medium size subject pool. Bryman (2012a) recalls how in *Social Research Methods* (Bryman, 2012b) he cited Warren's (2002) suggestion that the minimum number of interviews in qualitative research needs to be twenty (Bryman 2012b, p425). Becker (2012), on the other hand, resists quantifying saying that every experienced researcher knows there is no answer to the question of how many interviews is enough; 'The only possible answer is to have enough interviews to say what you think is true and not to say things you don't have that number for' (Becker, 2012, p15). He suggests that there is no universal 'right place' to stop and this decision is often quite arbitrary; 'probably more the result of running out of time or money or some similar mundane

consideration than of some logical analytic procedure' (Becker, 2012, p15). Denzin refers to the 'method of instances' when responding to this question. As long as analysis is uniquely adequate for the phenomenon, his answer is clear: 'How many interviews are enough? ONE' (Denzin, 2012, p23). Deciding the right number of participants to be included in a study depends on meeting the requirements of the epistemic community (Doucet, 2012), yet there are a variety of opinions about what that right number is. In looking at the epistemic community of the PhD study, other work has indicated that qualitative PhD studies commonly have sample sizes of between 20 and 30 participants (Mason, 2010). My own study involved 27 participants (26 parents) in 36 interviews, each lasting 30 to 240 minutes. The study sample was adequate to gain an in-depth understanding of the lived experience of parents.

### *The study design*

I have outlined how participants were recruited and described the methodologies informing the research. In this section I consider the design of the study looking at how data was collected and the design of the interview guides. The methodologies described in section 3.2 informed the kind of study I was carrying out; qualitative, semi-structured and to varying degrees in-depth, (inter) active, reflexive dyadic, narrative. The starting intention was to carry out only face-to-face interviews with participants, however I also carried out telephone interviews with parents who were not living in central Scotland. Methodological textbooks have suggested that the telephone mode is not best suited to the task of qualitative interviewing (Gillham, 2005; Rubin and Rubin, 1995), with development of rapport potentially affected by lack of face-to-face contact (Shuy, 2003; Gillham, 2005). However, telephone interviews can also be seen to allow the inclusion of participants across a wider geographical scale, the next best thing to being there (Tausig and Freeman, 1988) and may offer greater anonymity when the topic is sensitive and therefore reduce the risk of emotional harm (Kavanaugh and Ayres, 1998). There is also evidence that telephone interviews may be a good alternative to face-to-face approaches and may generate useful data and achieve successful interaction (Chapple, 1999), in some cases being described as 'unexpectedly rich' (Chapple, 1999, p91) and capable of achieving as good a rapport as face-to-face interviews and generating excellent data. While face-to-face interviews were the

principle methods of data collection, a small number of telephone interviews were carried out with parents it was not possible to interview in person.

The design of the interview guide (Appendix 2) reflected the methodological approach to the research and was also informed by previous research with families and other relevant literature. As described in chapter 2, key themes regarding the significance of diagnostic information were identified in research with families living without a diagnosis. These included: knowing the cause/etiology of their child's condition; information about the future/prognosis; acceptance; labels; and access to support and services. I was keen to explore whether these themes were relevant for participants in my own research. However, I did not want to force discussion of these themes, so I designed the interview guide in a way that allowed as much opportunity as possible for these themes to emerge spontaneously, if they were going to, prior to asking parents about these areas of research interest.

The interview guide was used in all first interviews with parents interviewed face-to-face (19 parents). Parents were invited to begin their interview by sharing their story. They were invited to begin their story wherever they wished: from before they had their child, when their child was born, or when their child started to experience difficulties. Most parents chose to start their story before their child was born, and spoke of the pregnancy. Some began their story even earlier, speaking about their own families or the hopes they had for starting a family before they conceived. Frank (2002) tells us, in his own experience of interviewing, that most people are eager to share their story. I found this to be true in my own research, and often the participants would talk for around 30 minutes in response to this first open-ended question prompting a narrative history. I didn't try to guide their narratives in a particular direction and used no directional prompts. Other than clarifying words or phrases and using continuers like 'mm', 'uhuh', 'yeah' (Schegloff, 1982) to support them in the telling of their stories, I didn't interrupt or direct the parents in the telling of their stories.

Following the telling of their stories, parents were asked several general questions to provide more information about their child's difficulties and their perceptions of the current situation regarding diagnosis. I then began the next section of the interview by asking parents a general open-ended question about whether they felt not having a diagnosis had

had an impact on their lives, practically or emotionally, and parents were encouraged to elaborate. Only after this did I ask specific questions about key areas identified by previous research. Parents were asked about these specific areas of research interest (Cause/Prognosis/How to describe their child's disability/Access to services/Access to support/Acceptance). I referred back to issues already raised in their narrative or responses. Many of these areas had already been mentioned by parents, so this gave further opportunity to clarify and for further discussion, as well as an opportunity to respond to questions about areas they had not already mentioned. I then worked through the rest of the interview guide with the parents, maintaining flexibility of structure by changing the order of the questions if the context of the interview required it.

With the hope of eliciting in-depth data about the meaning of diagnosis for parents, I asked them how they *felt* about not having a diagnosis; how often they thought about diagnosis; whether they used to think about it more; why they thought a diagnosis had not been made; how likely they thought diagnosis was; who they saw as responsible for diagnosing their child; how important they thought diagnosis was and what benefits would it have; whether there were any negative sides to having a diagnosis and whether they knew of other families of undiagnosed children or knew about undiagnosed disabilities prior to their experience with their child. They were also asked what, for them, was a diagnosis; and what wasn't. After this the process of diagnosis was considered and they were prompted to describe whether they had looked for a diagnosis and in what way they had done this. They were also prompted to talk about how they came to realize that diagnosis may not be possible. The final section of the interview explored their perceptions of doctors and medical science.

Several parents made reference to how the questions they were asked in the interview were very relevant. It is difficult to trace my decision-making regarding each individual question in the guide. Other research with families was influential in identifying key themes, although some of the other more abstract questions (like asking about their perceptions of medical science or what they thought a diagnosis was and was not) came from theoretical literature, the findings from my MSc dissertation, or even an intuitive sense of wondering about the possible meaning of absent diagnosis.

For the additional 8 parents living outside of central Scotland, an electronic version of the interview guide (appendix 4) was sent to them by e-mail to complete and send back. The questions asked, therefore, were the same for participants interviewed face-to-face and those completing the interview form. From their written responses, an in-depth telephone interview guide was devised much in the same way that second interview guides were created for other participants (an example of which is included in appendix 5). Completing the interview form was a time consuming task with many of the questions provoking in-depth responses. Yet the parents who expressed interest in taking part keenly completed the forms with very detailed responses. I then conducted a follow up telephone interview with these parents. Telephone interviews lasted between 60 and 180 minutes. I asked about issues they had raised in their interview form as well as additional areas of research interest they had not written about. Seven parents were included in the research in this way, 6 mothers and 1 father. All telephone interviews were audio-recorded and transcribed ad verbatim.

An additional face-to-face interview was carried out with the founder of the support organisation Syndrome Without a Name (SWAN) in the UK. This interview was also audio-recorded and transcribed ad verbatim. The interview explored her experience of setting up the support organisation for families living without a diagnosis and also her own personal experience of being grandmother to a child without a diagnosis. As with the other participants, consent was given for data from the interview, including direct quotes, to be included in the thesis and related documents; with the participant being aware that her role as founder of the support organisation meant that she was easily identifiable. Information about the support organisation SWAN UK and discussion of the interview with Liz, the founder of SWAN UK, is included in Appendix 7.

The design of the study allowed for the elicitation of rich, in-depth data. Interviews were listened to and transcribed prior to second interviews (if relevant) being carried out. The next section looks at how data was analysed, detailing both the process of producing transcripts from the interview data and of developing themes and analysing narratives.

### 3.5 Methods of data analysis

#### *Producing transcripts*

All but one interview was audio recorded with permission. One parent chose not have our interview audio recorded, so notes were taken instead. For this reason there are very few direct quotes of this parents' interview (Kamil's mother), however I describe important aspects of our interview in the narrative analysis in chapter 5. Stuckey (2014) acknowledges that by recording interviews, the interviewer can focus on listening and responding to the participant, without being distracted by needing to write extensive notes. I learned this in the long (5 hour) interview with Kamil's mother, in which note taking at times got in the way of our interaction and made it difficult to document direct quotes beyond a sentence or two.

Recorded interviews were transcribed ad verbatim; with transcribing done by myself. Developing a good quality ad verbatim transcripts of interviews is a critical component of the qualitative research process, as the language used by participants becomes primary data that helps us to understand behaviour, processes, and the cultural meanings of participant's perspectives (Hennink and Weber, 2013). Although not a conversational analysis study, I was keen to transcribe in a way that reflected the parents' narrative closely. I intended to transcribe all interviews fully; including word repetitions, partial words, pauses, silences, false starts, fillers and acknowledgements used by both myself and the participant. As acknowledged by other authors, this was extremely time and labour intensive (Kvale & Brinkmann, 2009).

There are mixed reviews about the benefits of transcribing at this level of detail. Sandelowski (1994) suggests that for most analytic qualitative approaches, it is not beneficial to transcribe fillers and that the transcription process needs to focus on the accuracy of the content. Seale and Silverman (1997), however, recommend the use of the transcription symbols of conversational analysis including the fillers and pauses in speech, asserting this enhances the data. MacLean, Meyer and Estable (2004), in their examination of the improvement of the accuracy of transcripts in qualitative research, found that including fillers was not only

unnecessary but at times counter-productive, interrupting the flow of the participant's dialogue and adding nothing to analysis. I transcribed the verbal component of all of the interviews I carried out fully and ad verbatim. I also noted pauses and silences and devised a notation system for inaudible sections, emotional content, and anything else happening in the background. For the purposes of presenting data in analysis chapters, however, I realised that my own conversational fillers made it difficult to read parent quotes. In direct quotes in data chapters I have represented pauses in parents' speech by leaving space between words, however I have also added in limited punctuation (full stops and commas) that I felt reflected the flow of the parents' dialogue as well as emphasis marked by italics where it was particularly noticeable. Transcribing fully allowed me to later decide which aspects of the transcribed dialogue added to meaning and which got in the way of understanding what the parent was saying, and so I could edit knowing the original form was preserved in the transcript. Once transcripts were complete, their accuracy was verified again by reading the transcript while listening to the audio recording. Transcripts were printed out so that I could note any observations when reading them over, and were stored securely in a locked filing cabinet in my secure University office. Participants were offered a copy of their transcript, which around a quarter requested. None of the parents raised any issues with the way their interview/s had been transcribed.

#### *Developing themes and narrative analysis*

Transcripts were printed out (with pseudonyms and other measures to ensure anonymity in place) and read through several times. Notes were made of general observations during these readings. Transcripts were converted into rich text formatting, so that they could be imported to an appropriate software package to assist data analysis. Qualitative Data Analysis Software (CAQDAS) enables researchers to manage qualitative data that would be too difficult using manual pen and paper methods and facilitates efficient data indexing and management. Themes were developed in two ways; either they were a priori themes identified in previous research and then identified (or not) in the transcripts, or they were grounded in the data and emerged throughout the process of data analysis. I used different methods of recording themes and analytical ideas. I made use of the software package



NUD\*IST for thematically organising transcripts and coding nodes, and took extensive notes for grouping themes and developing analysis. Appendix 6 lists the themes identified as free nodes using the NUD\*IST software, from which I developed sub-themes and developed analysis.

A priori themes were easier to code. Parents raised issues associated with these themes spontaneously (that is, without prompting) and also were asked specifically about these themes. In analysis of the data I created a node to differentiate between the issues raised spontaneously and those raised as a consequence of prompting. Themes that were not a priori, but emerged from the data, were identified throughout the process of analysis. This involved identifying a potential theme, coding it, and then returning to see if the theme was present in the other transcripts already coded. Therefore, there was an iterative process of coding and re-coding; I called this 'back-coding'. At times a theme would be identified in a transcript for the first time, for example 'being kept on at the clinic', and when returning to back-code the other transcripts it was then observed in other accounts. Iterative reflection on what was already coded was an important part of the analysis process.

In carrying out thematic analysis using NUD\*IST software, I created an audit trail of developing themes that I could refer back to if need be. A new version was saved once I felt there were significant enough developments in analysis to justify a new saved version. In the later stages of the project I converted the most recent version of the study to NVivo 10. By the end of the data analysis process there were 31 versions of the interview study on the NUD\*IST software. All electronic data generated from interview recordings, transcripts, qualitative software analysis and other notes were held securely on an encrypted computer drive.

In addition to using the software to enable coding and thus the organising of themes across the 36 interviews and additional interview documents, a crucial aspect of the development of themes and analysis was my critical self-reflection. Akin to the recording of memos on NVivo, I recorded my thoughts and ideas in a series of A4 writing books and also electronically in word documents. By the end of the thesis I had extensive written notes about all aspects of the project. The way that categories and related sub-categories were organised and related was therefore by a 'pen and paper' process. Whole nodes were

printed out and I then identified sub-themes within nodes. For example for the 'cause' node one of the sub-themes was 'risk of recurrence', which had further sub-themes including 'decided to have another child'. Associated analytical notes on this sub-theme may have included thoughts on how this decision was made, emotions described etc. CADQAS was crucial for the organisation of free nodes but the equivalent of the memo creation, and the tree structures of CADQAS to clarify ideas and identify relationships with other categories, was carried out on paper. I found this more practical and felt less restricted by the format of the software.

In carrying out the narrative analysis, I returned to the whole story of the participants, reading over their whole transcripts several times and making analytical notes. In considering the kind of stories parents told me, I identified sections or moments in their narratives when their story reflected a particular narrative typology, for example Frank's (1995) restitution, quest, and chaos narratives. In doing so I became aware that these typologies needed to be modified to reflect the truths of the parent accounts. I also coded for metaphors used by parents which involved close examination of every word and phrase they used throughout their accounts and creating a 'metaphor' node, which later I developed into subthemes, including 'stasis', 'battle' and 'puzzle' metaphors.

When considering how the data from each participant should be included in the analysis, I made a clear decision early on in the analysis process that data from all parents should be included in the analysis chapters. Although three of the parents (Tom's mother, Rupert's parents) are identified as having experiences atypical to that of the other participants, data from their interviews is included where relevant in data chapters. When their data has been excluded from analysis as the context of the section or chapter is not relevant to their experience, then I clearly highlight this. I also made the decision to include all parents together (26) when analysing the data, whether they were interviewed face-to-face as most parents were, or by telephone. The telephone interviews yielded rich and in-depth data and it was felt the experience of those parents were as relevant to the analysis as the parents interviewed in person. Perhaps the telephone format enabled the parents to share intimate details of the experiences and feelings easily, for sensitive topics were covered and I gained a deep understanding of their lived experience through collecting and analysing their interviews.

In referring to parents' narratives I refer to the whole story told to me across our research encounters. I was keen to respect the integrity of the parents' whole story. To contextualise direct quotes, however, I have noted further information after relevant quotes indicating method of data collection (i.e. telephone interview or interview form where participant was not interviewed face-to-face), which interview the quote is from (i.e. 1 or 2), and the country of residence of the parent (ie England, USA or Australia where the participant was not from Central Scotland). Those participants who were interviewed face-to-face (the majority of parents) lived in Central Scotland. I have not indicated method of interview or country of origin after the direct quotes from the interviews with these parents; therefore all quotes unless indicated otherwise are from face-to-face interviews with parents from Central Scotland. Where this is not the case, country of residence is noted (ENG, USA or AUS) and method of data collection, i.e. telephone interview (TI) or interview form (IF).

Following further personal reflection in 3.6, the stories parents told me of their experience of living without a diagnosis will now be told through analysis of the data in the following chapters.

### **3.6 Personal Reflections**

*As already described, my intention to carry out interviews in which mutual self-disclosure was a strong feature, and to tell my own story of living without a diagnosis in interludes in the thesis, changed over time. I learned early on in the interview study that in-depth self-disclosure did not seem appropriate in the interview setting. With reference to telling my own personal story in the thesis of parenting Maya, and later Willow, the task was much more difficult than anticipated. I have always been fiercely protective over their privacy, known among health, educational and social work professionals as the mother who will never sign a blanket consent for the sharing of information and refusing to have EYCAT (multi-disciplinary) meetings because I saw them as an act of intrusive surveillance. Instead, I acted as our family's own 'key worker', controlling the information shared between professionals and co-ordinating the girls' care. Yet in the thesis I was eager to share a story that told not only of the difficulties and challenges of parenting a disabled child, and further a child*

*who does not have a diagnosis, but also the joy, given that the joyful aspects of mothering a child with significant development delay have often been overlooked in research with families in favour of a reading of disability as tragedy (Kearney and Griffin, 2001). Yet, in the end, I did not feel comfortable telling this in-depth story and so my offerings are a small fraction of the story I have to tell. As Amy Boesky admits in 'The Story Within: Personal Essays on Genetics and Identity' (Boesky, 2013), once the personal story of living with a genetic condition or genetic mutation is published, you cannot take it back.*

*Over the years of the PhD I balanced the caring role with working as best as I could. As a single parent for the first four years, and with childcare very difficult to access due to Maya's mobility and medical needs, it inevitably took longer to progress. Lack of boundaries around what to expect from Maya's future, like associated health conditions, meant unexpected challenges that I had not anticipated. I had always been an anxious person, so mothering a baby who at times forgot to breathe came with its challenges. Luckily, she was a remarkably healthy child, despite her physical disability. She ate orally from the age of 2 and used a walker and wheelchair for mobility from the age of 3. She could speak long sentences before she could move, and was articulate, sociable and wise. She asked questions as a young child that would provoke me to think and reflect, and that could spark a heartfelt sorrow as easily as a grateful joy.*

*Childhood muscle disorders are often accompanied by associated health conditions like respiratory and cardiology problems, thankfully she had neither. Neuromuscular scoliosis is one disease often associated with muscle disorder. Not knowing this, it was a shock in 2006 to learn her spinal curve was progressing rapidly and that she would need a spinal brace (worn 23 hours a day) and major corrective surgery to stop her scoliosis becoming life-threatening. There's nothing that quite challenges a usually optimistic person as a progressive disease. Sometimes I would watch Maya playing on the floor and think 'what have I done to you?' imagining the possibility of Maya being herself but without the disability. In the same way that Landsman describes asking herself 'might my DJ now be normal?' if she had acted differently (Landsman, 2009), I was sometimes haunted by a self-blame that lacked evidence or specificity. These thoughts were fleeting, life was too busy to hold them in the foreground. At this time, I still thought that something in pregnancy or labour had caused her disability.*

*Matthew, my partner, moved in after finishing his PhD in Nottingham and we got married when Maya was 7. I started to anticipate Project Baby. Keen to have two more children, and looking forward to a completely non-medicalised experience, I was excited. My PhD was progressing well, despite the significant disruption the caring role had on consistence attendance. Maya and I had an on-going joke at that time. She knew I didn't have strong religious beliefs, although I'd always encouraged her to make her own mind up about the existence, or not, of god. I would tell her that when queuing for children I had definitely picked the right queue, and she would reply that I didn't believe in queues.*

*On becoming pregnant with Willow, early in 2009, I began to prepare for an experience that would heal the hurt of the journey I had been through with Maya in her early months. I planned a home birth with the support of local midwives and at 18 weeks pregnant I trained as a doula, planning to also train in hypno-birthing and to use the techniques for my own labour. My thesis was due to be submitted two weeks before Willow was due in October 2009. It was progressing well, although I eagerly anticipated the publication of Jutel's (2009) outline for a sociology of diagnosis, expecting that my work would finally have a sociological home and that I could incorporate this work into my final draft.*

*About half way through the pregnancy, Willow stopped moving. In the end, I spent much of the last part of my pregnancy in hospital. The consultant neonatologist and my obstetrician were initially confused by the apparently severe difficulties the baby was having; as she was unable to swallow the amniotic fluid was rising and premature labour was imminent. As I didn't present with any apparent muscle weakness, it being a dominantly inherited condition was initially ruled out and it being a recessively inherited condition repeating with different parents was in the realm of 'fairytale statistics', in the words of the neonatologist. It was only when the regional geneticist was brought in for consultation that the possibility of it being the same condition were realised.*

*My muscle strength was tested and I found myself narrating a history that suddenly rewrote itself as it was told; my 'W sitting' as a child, my hyper-flexibility, that I was never very co-ordinated at highland dancing or swimming, my painful skinniness as a child, the pronated feet that needed built up shoes, my clumsiness. These aspects of myself were suddenly significant, collated and viewed through the lens of genetic disorder. There was a focus on my running. Did I tire easily? I never*

*thought my half marathon time would qualify me for anything, let alone a genetic condition. Personality became pathology overnight, no not overnight, in a moment.*

*To cut a very long story short, another year and a trip to the Newcastle Life Centre later, it was decided that the best guess was that I had a de novo condition, occurring in me as a very young embryo, that I only had a very mild version of. A weakness was found in my upper arms that wasn't consistent with the strength in the rest of my body. I suddenly had a reason for never having been very good at press-ups! As Dimond (2014a) describes, the extension of the clinical gaze includes the parents' body; my physical features were collected as I was collaged by the clinic. I now had a new genetic identity.*

*Willow was born 2 months early in August 2009 and was ventilated for a month. She came home at 6 months old with a gastrostomy, tracheostomy and, after much pushing on my part, a team of NHS night nurses who took care of her overnight at home. She needed suction around 100 times a day. Parenting a child with a tracheostomy was more than a challenge to my anxiety; when even dandelion clocks are fraught with risk, and sand and water are no longer playthings of the child but a potential threat to life. There is no literature looking at guilt or self-blame when a parent has a de novo condition that doesn't present strongly in them but causes significant impairment for their child. My 'molecular oddity' (McLaughlin and Clavering, 2011) haunted me. For Maya to understand why her sister had the same condition, I used the metaphor of a wonky brick. The mutated gene was from then on referred to in our home as 'mummy's wonky brick'.*

*Like her sister, Willow was a healthy child. It was the mechanics of her body that caused her difficulties; she wasn't particularly prone to illness. As I was obsessive about tracheostomy tube infection, however, I avoided socialising. We made up for the loss of the private bond of the early months with days of me holding her, feeling gratitude that she had survived. The thesis was put on hold for near enough 2 years, during which the sociology of diagnosis emerged through key publications in 2009 and 2011. Willow slowly developed, and it began to look like she would one day be strong enough to manage without her tracheostomy. She became a cheeky, stubborn, delightful toddler; entirely reliant on our care to keep her safe. In the meantime, the dialogue with genetics was ongoing. New tests were carried out in the search for diagnosis. A conversation began about the reasons for continuing to pursue diagnosis. We were keen in the future to have a child who didn't have the same condition as the girls. The geneticist talked through the process of PGD IVF. Based on*

*the remote chance that we could locate a genetic diagnosis, the process of IVF was risky and not guaranteed to be successful. She suggested a test to assess my fertility levels, given I was 36.*

*The results of the fertility tests, received in July 2010, were shocking and apparently conclusive. My AMH hormone levels were so low that fertility was described as 'undetectable'. I came off the pill after learning of my infertility, keen to lower the risks associated with long term hormone use. Following a consultation at the menopause clinic in early October 2010, I was diagnosed with early menopause and confirmation was given in writing that, even should a diagnosis be found, I would not be eligible for IVF.*

*It was a sense of closure. My girls brought me joy and I was grateful for them. I focused again on my thesis. Absent diagnosis was, ironically, both the catalyst to the thesis and the obstacle to its progress. With reference to reproductive decision-making, in a sense I 'choose not to choose' (Kelly, 2009) in the face of carrying a genetic mutation that had devastating consequences for my children. The risk of recurrence, if it was a dominantly inherited condition as suspected, was 1 in 2. To have been allowed to escape the culpability of choosing to choose, really was what I wished for. But I came to terms with being a different kind of mummy and my girls inspired and amazed me.*

*The new date of submission for my thesis was the end of September 2011. In November 2010, I discovered I was pregnant.*

### **3.7 Conclusion**

In Chapter 10, I will continue to tell my story. Here, I have shared some of my journey, keen to reciprocate after hearing the stories of others, yet at the same time holding much of this story back to protect the privacy of my family. Ruth Behar says about the researcher and the researched: 'we ask for revelations from others, but we reveal little or nothing about ourselves; we make others vulnerable but we ourselves remain invulnerable' (Ruth and Behar, 1996, p273). In offering a version of my story, I have made myself vulnerable and, it is hoped, given something back.

This chapter has explored the methodologies informing the data collection, ethical considerations of the study, the research design and methods, and the methods informing data analysis. While the journey to find the right methodologies for the data collection took place early on in the study, all other aspects of the process of considering methods and methodology has been on-going, shape shifting with each iteration to the literature and as the study progressed. In qualitative research the design should be flexible rather than fixed (Robson, 2011); a reflexive process working through every stage of the project (Hammersely and Atkinson, 1995 p24). This chapter has laid out methods and methodologies, and has described the process of deciding on methods and methodology as a journey. I have also shared something of my own journey.

In the following chapters, I turn to the stories of the parent participants. Analysis of these stories, both thematic and narrative, forms the next six chapters looking at the process of diagnosis, parent narratives, and the consequences of living with absent diagnosis. Chapter 4 examines at the parents' quest for diagnosis, and is therefore concerned with the process of diagnosis. Chapter 5 explores the narratives of the parents, the telling of non-diagnosis. Chapters 6, 7, 8, and 9 focus on the perceived consequences of absent diagnosis by looking at how parents made sense of living without a diagnosis and the impact of not having a diagnosis on how they thought about cause and prognosis and on access to services, support and in managing social interaction.









## **Chapter 4      The process of diagnosis: the parents' quest**

### **4.1      Introduction**

### **4.2      How parents were agentive in the process of diagnosis**

Use of the internet to seek diagnosis

Difficulties with online searching

Parents seeking others to assist with the search for diagnosis

Parents looking for other children like their own

### **4.3      How the search for diagnosis changed over time**

Triggers for increasing or resuming the search for diagnosis

Factors contributing, reducing or stopping search activity

### **4.4      How parents made sense of the diagnosis process**

Parents' understanding of genetic testing

How parents made sense of possible genetic cause

Parents' perceptions of genetics as an advancing field

### **4.5      Conclusion**

## 4.1 Introduction

This chapter is the first of six chapters analysing the data from the interview study (chapters 4 – 9). In this chapter, I look at the parents' quest for diagnosis and other aspects of the process of diagnosis. I consider how parents were agentic in the search for diagnosis, how the search for diagnosis changed over time, and how parents made sense of the testing process.

As already described, diagnosis has different meanings. Drawing on the work of Blaxter (1978), and the more recent work on the sociology of diagnosis (Jutel 2009; Jutel and Nettleton 2011), we can conceptualise diagnosis as *process*, as *consequence* and as *category*. In this chapter, I consider the process of diagnosis for parents, in looking at their quest for diagnosis and how they made sense of that process. A *process*, depending on context, can be defined as:

A series of actions or steps taken in order to achieve a particular end  
A (natural) series of changes  
(Oxford Dictionary, Oxford University Press, 2010)

A forward movement  
The course of time  
(Collins Dictionary of Sociology, 2005).

Here, I suggest that these multiplex dimensions of process are apparent in parent narratives. The process of diagnosis for parents involved a series of actions in order to achieve a particular end, change, a sense of moving forward towards the telos of diagnosis, and was influenced by the course of time. The parents' journeys involve the multidimensional concept of process. In being agentic in the quest for diagnosis and in their sense-making activity, process in the sense of a series of actions or steps taken in order to achieve a particular end was enacted by the parents. The process also represented change over time as search activity and focus on diagnosis fluctuated and ultimately reduced over time. Despite a common theme of reduction in search activity over time, the process of diagnosis for parents of undiagnosed children may be unending, albeit decreasing in intensity. The process lacks closure; it continues, ebbs and flows, without the end result of diagnosis being realised. Perpetuity can be seen as a unique characteristic of the diagnosis process for

parents of undiagnosed children. Although the felt need for diagnosis may lessen, parents may remain alert to new diagnostic opportunities and the quest for diagnosis remains unresolved.

As parents commonly experienced the seeking of a diagnosis as an intense, prolonged event, I refer to their *quest* for diagnosis. A quest is a journey towards a goal which, in mythology and literature, usually sees the protagonist overcoming obstacles and travelling far to achieve what they seek. In other literature looking at the intense seeking of a diagnosis by parents, the diagnosis process has been described as a 'quest' for diagnosis (Rosenthal, Biesecker and Biesecker, 2001; Maher, 2013; Graungard and Skov, 2007; Watson, 2008, 2009); a 'diagnostic odyssey' (Reiff et al., 2012).

The beginning of the process of diagnosis for parents of children with development delay is realising or acknowledging that their child has difficulties. The process of diagnosis for some parents began *before* they had contact with medical professionals, when they noticed something about their child that was different from the norm in terms of their development, behaviour or health. There may have been a delay in the health professional/s agreeing that their child had a significant difficulty. For other parents it may have been the other way around, with the health professionals involved noticing a difficulty and parents taking time to accept that their child had this difficulty. Some parents instigated the diagnosis process, while others joined in the process once they acknowledged that something was wrong.

This chapter explores the parents' engagement in the process of diagnosis. In section 4.2, I explore their agency by considering: parents' use of the internet to seek diagnosis; difficulties with online searching; parents seeking others to assist with the search for diagnosis; and parents looking for other children who resembled their own. In 4.3, I consider how the search for diagnosis changed over time by looking at triggers for increasing or resuming the search for diagnosis, and factors contributing to the search for diagnosis reducing or stopping. In 4.4, I examine how parents made sense of the testing process in the context of genetic testing, examining: parents' perceptions of certainty and uncertainty in genetic diagnosis; how parents made sense of possible genetic cause; and parent perceptions of genetics as an advancing field.

## 4.2 How parents were agentive in the process of diagnosis

Parents were not merely passive receptors of diagnostic information; they were active in the search for diagnosis, influencing how the process of diagnosis was shaped. There was variation in how involved each parent was in this process, but commonly parents contributed momentum to the search for diagnosis, urging clinicians and others to seek diagnosis, and were also active in their own diagnosis work by exploring symptoms, seeking new avenues for possible diagnoses, researching suggested or tentative diagnoses, and actively seeking others to help locate diagnosis. Knowledge the parents obtained in their search for diagnosis was commonly fed back to clinicians as the iterative process of diagnosis evolved.

Parents conducted multi-method searches in their quest to obtain a diagnosis for their child. They consulted books, leaflets and other publications ('We got books from the library' Calum's mother); accessed information via newspapers, the radio or television ('If there's something on telly you're looking and you're thinking oh that sounds like that I wonder what he's got and you're just trying to put a label on it yourself' Calum's father); consulted with doctors, consultants and other specialists ('I've been in touch with a lot of doctors trying to find out what is wrong with him' Ismail's mother); consulted other parents, friends or family members ('We also had a friend do a psychology assessment on him' Peter's mother); attended conferences and relevant events ('I went to a conference and during a diagnosis session it was decided she didn't have that syndrome' Anna's mother); and, most commonly, accessed information on the internet ('I was looking for an answer really on the internet' Tom's mother). The internet was cited as the most frequently used tool for searching for a diagnosis, although all parents described using multiple methods to seek a diagnosis.

We've looked on the internet, have read paediatric text books, read pamphlets about certain syndromes, talked with other parents and health professionals. We've taken our son to numerous tests in the hopes of finding a diagnosis (Peter's mother, IF, AUS)

The small number of parents (five) who reported that they did not conduct extensive internet searches for a diagnosis either did not have internet access at home when their child was young (two parents) or described how their partner searched online for a diagnosis and shared the results of their searching with them (three parents).

*Use of the internet for self-diagnosis and to explore suggested diagnoses*

As well as using the internet to find out information about individual symptoms and characteristics of their child's condition, parents commonly sought to locate a specific diagnosis, that is, to self-diagnose their child's condition.

You kind of try and self-diagnose don't you, you go through everything and you think oh well he ticks that box (Calum's mother, 1)

You're just trying to put a label on it yourself (Calum's father, 1)

In trying to self-diagnose their child's disability, parents carried out a process of alignment and exclusion (Latimer et al., 2006), seeking to match their child's signs and symptoms with conditions they read about online and excluding conditions that did not seem relevant.

I've spent masses of time on the internet, researching characteristics that she has and trying to match them with syndromes (Jessica's mother, IF, AUS)

Parents commonly conducted Google searches as a starting point to identifying conditions their child might have, using their child's characteristics or medical signs and symptoms as keywords in their search for a unifying diagnosis.

I do Google searches and literally put in every single one of her phenotypical characteristics or symptoms like I'd do...Google searches on developmental delay and you know eh a wide distance between the nose and pronounced epicanthic folds and it's amazing how much you'd come up with and then I'd start trawling through all these syndromes (Eva's mother, TI, ENG)



When we initially found out he had hypotonia we were on the web you know putting in hypotonia (Abdul's mother, 1)

This search activity often yielded vast amounts of information, which parents had to manage and sort through to find information that may be relevant to their diagnosis search, excluding that which did not seem relevant. Parents had difficulties fitting their child into a diagnosis or syndrome using online resources: 'I'm coming up with all these syndromes and none of them quite fit' (Eva's mother, TI). As Claire's mother describes, she couldn't identify a condition that completely fitted with all of Claire's difficulties.

Cause we've looked up all what her problems is on the comp on the internet either in books and everything we've tried everything and nothing comes up that's got all her problems some of her problems yeah but not all (Claire's mother, 1)

Using their child's signs and symptoms as clues, parents carried out some of the detective work of diagnosis. They had difficulty locating conditions that fitted well with their child's own condition, and none of the parents managed to identify a specific diagnosis using this practice of 'lay diagnosis' (Beach, 2001). In ruling out diagnoses, parents interpreted and managed the information available to them. They were also able to uncover new possible avenues for exploration and take back to the meetings with their child's consultant suggestions or questions about specific diagnoses.

Researching diagnoses parents were speculating about was also a key research activity for parents and they actively followed up diagnoses that had been suggested to them from a variety of sources, including their child's doctor or other parents, friends or family.

I trawl I mean the minute the doctor said oh we're testing for Fragile X I went oh Fragile X website you know reading it (David's mother, 1)

Other people suggest things to you and you go off with that...we've been down the line of autism we've been down the line of Angelman's syndrome been down the line of Fragile x...and the speech therapist then suggests Auditory Processing Disorder so you go down the line of that people suggest things and you deu deu deu let's go and look at all this (Emily's mother, 2)

The search process was therefore an iterative process involving a to and fro between resources parents encountered through their own search activity and through the diagnosis

process of the clinic. Parents considered multiple possible diagnoses in their diagnosis journey, potentially having more than one suggested or tentative diagnosis at a time that they were contemplating and researching to determine how relevant it may be.

It was very much a case of let's read everything in sight...you could see wee traits of Cameron and you know there was times where I maybe read something about autism and I used to think that's what it is, he's got autism, and then I heard of somebody that had Asperger's and I read about Asperger's and I thought well I can see Cameron there...maybe you read about something else you think well maybe he could fit into that category as well, maybe no fully but he could you know? (Cameron's mother, 1)

In some cases, diagnoses that had been suggested to parents by their child's health care team were ruled out by the parents when they researched the conditions themselves. Claire's mother, for example, described a process in which herself and her partner had been given a tentative diagnosis for their daughter. When they began to research the diagnosis, however, they discovered that it didn't 'add up'. They took their queries regarding the diagnosis back to the consultant, who agreed that the diagnosis wasn't accurate.

Me and [partner] done some study ourselves went through the computer eh me and [daughter] went to the library with all the names and all that and it just didnae add up cause the [clinical symptom] that's what she had but she had other things as well which didnae add up, and that's when we spoke to Doctor [name] and that's when he said he agreed with us. He said he wasn'y happy with it either so he must have obviously been discussing it as well (Claire's mother, 2)

Later in the interview Claire's mother described how her partner, following their research, 'had a feeling' Claire didn't have the suggested syndrome. The doubt that the parents had following their own research work was confirmed in a later consultation with their child's geneticist.

Dr [name] actually said we're no happy with her being in that circle...but he was classing it as that at the moment (Claire's mother, 2)

In this way, the parents had gained enough knowledge to have an experience of knowing that the diagnosis wasn't right for their daughter.

Calum's mother similarly described a process of researching, and consequently having doubts about, a diagnosis suggested by her son's consultant.

I've looked them up. I was going through a wee sort of check list in my head oh well yeah he has that has that has that oh no you know and then I'd find something that he didn't have and I would sort of mmm I wonder you know? And that would put sort of doubts in your head thinking I wonder if they're on the right lines (Calum's mother, 1)

As with Claire's mother, her research led her to question whether the clinicians involved in her child's care were considering the right diagnosis, as her son didn't seem to fit with what she learned about the suggested diagnosis. In this way parents showed agency not only in carrying out their own search activity and also in challenging doctors' suggested diagnoses.

Parents journeyed through a process of exploring and considering each possible diagnosis, seeing if they could fit their child into that category, then possibly ruling it out or keeping it on as a prospective diagnosis. One parent used an analogy of climbing a hill to describe this experience of considering multiple diagnoses.

You climb a hill when you read about something and you think that could be Cameron. You climb this hill and you think we're going to get there we're going to get there and you get to the top of the hill and somebody says no that's no what it is and you come right tumbling right back down again and then you read about something else and then you start to climb the hill again...it was a case of you know well okay it's maybe no that but maybe it could be this (Cameron's mother, 1)

Cameron's mother's analogy expresses the work and risk of the process of considering multiple possible diagnoses, a metaphor for the difficulties of moving forward for parents on their diagnosis journey. Considering multiple diagnoses to account for their child's developmental disability can be an 'emotional roller coaster' for parents (Watson, 2009). The parent may invest emotionally in each potential diagnosis, considering the emotional and practical implications of each diagnosis. Different diagnoses may come with different prognoses, associated health issues, developmental difficulties, care needs, even life expectancy, and in considering each one the parent may begin to anticipate their child having that diagnosis. This is a stressor unique to the experience of the diagnosis process, and when a diagnosis process is prolonged this stressor may be more significant.

In using the internet to seek diagnosis or research suggested diagnosis, several parents talked about how their searching techniques had changed over time. Moy's Father, for example, described how initially he used to 'Google' general terminology, diagnoses or clinical features of his daughter's disability. Over time he used more specific terminology and different kinds of web pages as his knowledge increased.

I have still been using the internet but most of my searches have become more defined and from different sites. I now spend more time in clinical reports and medical journals than any associations or parent web pages (Moy's father, IF, USA)

As a father with no previous medical training, his experience of seeking a diagnosis for his child involves a process of proto-professionalization (Shaw, 2002, p289) in which his knowledge and use of medical terminology improves as he journeys through researching childhood disability. Similarly, Abdul's mother describes a change in the way she searches over time, while she still used the same 'tools' to search for diagnosis, she is increasingly selective about the terminology she uses when carrying out searches; 'I think it's more our understanding that has increased, we still use the same tools' (Abdul's mother, 1). In the process of searching online for a diagnosis, parents are likely to expand their knowledge about childhood disability and become more expert in their approach to the search process.

#### *Difficulties with online searching*

Around half of parents described difficulties with searching online for information that may lead them to a unifying diagnosis; the most common difficulties were being upset or confused by information they encountered online. Several parents described a cautious approach to accessing information online. Abdul's father, for example, was particularly careful when it came to accessing information available on the internet, and said: 'The trouble with the internet is it's full of junk and you know there's a limit to how much valuable information or useful information's actually out there' (2). Similarly, Calum's father described the internet as 'your worst enemy as well as your best friend' (1) and talked of the way it was 'filled with information but also filled with misinformation' (1). As Calum's mother also recognised, 'anyone can put what they like on the internet so you have to be careful' (1).

Claire's mother recalled being confused by the information she had accessed online. To make sense of this information, she had consulted books in her local library.

We got some information off the internet but a lot of it was like doctor-words so we went to, me and my daughter went to the library and got a book out there and read what the actual words meant (1)

Eva's mother, who had medical knowledge and experience through work, used the PubMed search engine to access medical journal articles in her search for diagnosis for her daughter. Despite her knowledge and experience, she still found seeking a diagnosis online 'mind boggling'. She found it particularly difficult to put the information she accessed into context.

I have no idea I can't put this into context...I'm coming up with all these syndromes and things and none of them quite fitted...I was finding it very difficult to put some of these things into perspective...I did find it mind boggling and confusing...sometimes I would find something and I would follow it through and get an article and then I'd think this is it this is it and then I'd read a bit more and think well actually I'm not sure (Eva's mother, TI, ENG)

Here, Eva's mother has difficulty finding a syndrome that 'fits' and, like Cameron's mother climbing her hill towards diagnosis, she experiences false summits of believing diagnosis may be near only for doubt to set in as she researches further.

Eva's mother has a background in animal science, so is familiar with medical knowledge in a different context. In describing the process of diagnosis, she refers to the difficulty she has sorting through and interpreting the information she finds online. She spoke of needing the help of an expert; 'a wise old man who knows a lot about this' (TI), to put the diagnostic information into perspective. Without this expert input; 'I needed somebody, somebody with perspective, somebody with experience who really knew about it' (TI), she struggles with the process of seeking a diagnosis for her daughter using online resources, despite using specialist search tools. Even for parents with education and training in a medical field, an expert in the specific field may still be needed to interpret and assist in the process of seeking diagnosis. Eva's mother is an active player in the diagnosis process; driving the process forward by urging her child's clinician to keep on looking, and researching new and suggested avenues for diagnosis taking the results of the research back to the clinic for

further negotiation. Like other parents, she carries out a lot of interpretive work herself. Yet she also needs the expert input of the consultant, in Eva's case the geneticist, to fully interpret the information she accesses online.

Along with difficulties understanding information accessed online, or not having the specialist interpretation skills to make full sense of the information, parents also described emotional difficulties associated with online searching for a diagnosis. Olivia's mother highlights the potential for the search process to cause upset and emotional stress.

You're frantically, okay I'll look up this and I'll look up that, what does that mean...I was getting myself so upset and stressed out because I was looking up all these different words all these different learning difficulties I'd never heard of before in my life...it was making things worse because you'd maybe start to look at things and think that she was a lot worse than she actually was (Olivia's mother, 1)

She comes across medical terminology she hasn't encountered before and imagines her daughter having the various syndromes, signs and symptoms she encounters online. Not being able to rule out the conditions she comes across online causes anxiety and upset as she worries about what the future might hold for her daughter. Olivia's father describes a similar process of becoming worried about the symptoms of syndromes he encounters online; 'the more you were reading in to it the more you were expecting fits to start or something' (1). He said he found it easy to 'jump to conclusions' about the implications for his daughter's future and acknowledged these conclusions did not always add up; 'you're sometimes putting two and two together and getting five' (1). For parents researching possible diagnoses online, there is a risk that upset may be caused by being unable to rule out specific conditions or assess the relevance of signs and symptoms. As there is an emotional element to the process of diagnosis for parents, putting two and two together and getting five may cause upset and worry.

Another way that parents were agentive in the process of diagnosis was in the seeking out of others to help them in their search for diagnosis. Several parents described actively seeking to make contact with relevant others who may be able to help them locate a diagnosis, including relevant health professionals as well as other parents of disabled children. They sought others online as well as in other ways, for example attending events or speaking to other parents or health professionals. Olivia's father, for example, described how he and his wife were planning to attend a conference in the hope of meeting someone who may be able to shed some light on his daughter's diagnosis.

We're going to a conference in [name of city]...I think that we're again just looking to I don't know we may stumble across somebody from somewhere or other that knows of or has seen this or I don't know but again it may be a grasping at straws thing (Olivia's father, 1)

Eva's mother, as described earlier, felt that she needed someone with more expertise ('a wise old man who knows a lot about this' TI) to assist her in her search for diagnosis. To illustrate the value of finding the right person to help in the search for diagnosis, Eva's mother tells a story of an acquaintance who sought a diagnosis for his own daughter. Her friend found the right person to help him in his quest and his daughter was diagnosed.

When he [the father] started to suspect she had Rett's syndrome he looked up who he considered was the best person to see and he had travelled with his daughter to see this consultant...he said that had been great for him because...he knew she was the right person...she diagnosed her and she gave him all the information and he went away feeling he knew what he was dealing with (Eva's mother, TI, ENG)

However, as Eva's mother described, in the absence of a diagnosis it was not possible to know who the right person to get in touch with was. Without a diagnosis, there was no person known to be expert in the field as there may be if a parent is faced with a strong likelihood of a specific diagnosis.

But when you've got a child with no diagnosis you think well who do I go to? You know I would do that but you think well, where is that? (Eva's mother, TI, ENG)

Other parents described making contact (online and face-to-face) with others in the hope of finding out more information that may lead to diagnosis. Contact was made with other parents or carers and also professionals or specialists in different contexts.

I go along to a parents support group and you know someone says what about this what about that have you spoken to this person, I think well give me their number and you know I'll sort of phone them or whatever (Cameron's mother, 1)

I do try and go out and try and find out or I'll go to events that are being held for parents and carers about certain illnesses and try and find out...also just speaking to other parents (Ismail's mother, 1)

I discovered through the internet another lady who is actually an audiologist...she's a community paediatrician but she specializes I think in audiology and I e-mailed her...I still keep in contact with her (Emily's mother, 1)

While few found their 'wise old man' (Eva's mother) who could help lead them down the right path to diagnosis, some made apparently useful contacts with others who could provide some clues in their search for diagnosis.

#### *Parents looking for other children like their own*

Another way that parents actively sought others to assist in the search for diagnosis was in searching for other disabled children who resembled their own child. Most often, this practice of diagnosis involved seeking to match clinical symptoms and features of their own and other children by looking through photographic images of other children online in an effort to find resemblance. This work of diagnosis for parents involved them acting to visualise similarity and difference between their own child and other children they met or saw images of online. Claire's mother described this process of seeking to find other children like Claire by reading descriptions of other children's difficulties. She thought she may have identified a possible diagnosis for her daughter when reading about another child in a magazine. However, after finding out more about the syndrome online, she realised they only had one characteristic in common so she ruled out the diagnosis.



It was a wee girl something to do with a growth disorder [syndrome name] but she had a couple of things I recognised with her like her pinky sort of goes into the ring finger...they had a website so I looked it up but the pinky was the only only thing I recognised cause if they said if they said like [symptom] and an [symptom] then I'd have clipped onto it and found out more (Claire's mother, 2)

Claire's mother also described how her family had constructed a website for her in an effort to locate diagnosis. By including images of her daughter on the website, it was hoped that others with children with similar characteristics ('just to see if anybody's got any similarities', 2) would make contact. The photograph was used as a tool to try to locate a diagnosis for Claire, with the family hoping that someone 'recognised' her.

Peter's mother described how she read the stories of the other parents on the support organisation SWAN (Syndrome Without a Name) website in the hope that she might come across another child like her son, that she might 'see someone similar' (IF, AUS). Jessica's mother, who worked with disabled children, described how she compared other children she worked with to her daughter in an effort to find similarities and hopefully a diagnosis. She also described how she looked for other children who resembled her daughter.

I'm always looking around...as parents we tend to gravitate towards people who are like us you know like us ...when I'm out and about...I do it all the time...I look at other children particularly if I see other kids in wheelchairs or who look disabled I look for similarities because you know I almost think that one day you know if I saw somebody that looked like Jessica I'd probably go up to them and say excuse me just to be able to say ah yes they're like me or yes that's what I experience (...) but there isn't anybody who knows what it's like to *be* us (...) I kept comparing the children I was working with with Jessica, and every syndrome I heard of I wondered if Jessica also had this (Jessica's mother, TI, AUS)

In this way Jessica's mother sought to recognise features or 'the look' of her own child when encountering other disabled children. The purpose of seeking other families with a child who resembled her own daughter was driven by the quest to find a diagnosis and also the desire for connection and solidarity with others. Moy's father was also keen to locate another child who looked like his daughter and who had similar characteristics. He regularly read a parenting magazine for parents of disabled children, describing how he compared pictures of the children featured in the magazine with his daughter to try and establish whether they shared similar 'traits'. Encountering visual images and clinical information of other children

could trigger his interest in and thinking about diagnosis; 'but then you see that picture of that kid in Exceptional Parent magazine and you think...hey Moy has a lot of those traits and my mind is off and running again' (IF, USA). He had a preference for websites and magazines with photographs so he could visually compare the children with his daughter.

When I do internet searches or when I read magazines like the Exceptional Parent magazine I prefer a lot of pictures like if it's a family website that actually shows pictures of a kid that does look different I really study them...I spend a lot of time searching for...Moy's physical symptoms (Moy's father, TI, USA)

Moy's father described how he was about to explore a diagnosis he had come across when looking at pictures and reading the description of another child. As this diagnosis had not yet been ruled out by him, it was described as one he 'still had' (TI); he was yet to explore with the consultant whether his daughter fitted with the diagnosis after seeing another child who looked like his daughter.

That's one that I still have and that one came from a picture of a kid from the Exceptional Parent magazine. In the syndrome the face, the eyes, the nose, you know sometimes you look at Moy and you think yeah she's a Down's baby and then other times you look at her and you say no not even close and so this kid looked more like Moy than any other kid so and we haven't followed up yet (Moy's father, TI, USA)

Looking for a child who resembled his daughter was one of the ways he actively sought diagnosis. He said; 'it's just whipping through magazines and seeing pictures and just staring into this kids eyes and going well yeah that looks familiar' (TI) In this way he was actively searching for children who resembled Moy.

Similarly, Eva's mother described how she sought diagnosis on the internet by looking at photographs of children with named syndromes to see if they looked similar to her daughter. Like Moy's father, who 'stared into this kids eyes' and sought familiarity, Eva's mother also looked at photographs of other children and tried to find resemblance.

I'd start trawling through all these syndromes and going to all these websites of children with these different syndromes and looking at photographs and thinking is that Eva? Could that be Eva? (Eva's mother, TI, ENG)

Eva's mother also posted a description of her daughter's signs and symptoms on a website for families with undiagnosed syndromes. On this website she also regularly searched the posts of other parents, looking for a childlike Eva. She had been in touch with several other parents in the hope of finding someone with 'exactly the same set of symptoms' as her daughter. She said;

I often look through because I'm thinking is there a description of Eva in here, has somebody else got an Eva out there? (TI, ENG)

None of the families at the time of the interview study had managed to locate a diagnosis by making contact with other families in this way, although such contact may have had other benefits.

In describing the different ways in which parents engaged actively and iteratively in a process of diagnostic searching, this analysis reveals two key features of their agency. Firstly, parents sought out and interpreted a diverse and complex range of information; secondly, they took their findings and suggestions to the clinic, challenging and engaging with professional diagnostic expertise. There was an emotional element to their engagement in the process of diagnosis. Also, although parents carried out a lot of interpretation work themselves, expert knowledge was still needed to fully make sense of the information they encountered. Parents searched for others to help them in their quest for diagnosis; seeking other parents or professionals who might have knowledge to help them with their search, and looking for other children who resembled their own.

#### **4.3 How the search for diagnosis changed over time**

The passing of time had an impact on the activity of searching for a diagnosis for parents. Commonly, the parents' quest for diagnosis decreased over time. Here, I identify triggers that increased, maintained or resumed parents' searching activity, as well as factors that caused parents to reduce or stop actively seeking a diagnosis.

Several triggers to increasing, maintaining or resuming the active search for diagnosis were described by parents, including: suggested diagnoses, routine appointments, and a change in the health, development or behaviour of their child.

As described above, parents actively researched diagnoses suggested to them by others. One trigger to resuming or increasing the search for diagnosis reported by parents was when friends, family or health/education professionals suggested diagnoses they had not previously considered or had not considered for a long time. Suggestions of diagnoses could trigger an increase in search activity for the parent or encourage the parent to maintain a search for diagnosis.

Speculation by people about syndromes has kept me searching (Moy's father, IF, USA)

Andrew and Lee's mother had more or less stopped her search for diagnosis as she felt she had exhausted all the possibilities for diagnosis. However, following a conversation about Fragile X and Angelman's syndrome with other parents while her children attended a disability sports club, she picked up her search for diagnosis again and returned to see the consultant about these syndromes as possible diagnoses. Even when the search for diagnosis has been dormant for a while, the possibility of it becoming active again may always be there.

Information encountered in leaflets, newspapers, on television or on the radio could also act as a trigger to resuming search activity.

What makes me start searching for a diagnosis are comments from people who say things like 'I know someone who is just like that, they have blah, blah' also TV programmes or documentaries (Jessica's mother, IF, AUS)

I listen to the radio sometimes, Radio 2 sometimes detox and stuff...there's been a few quite good telly programmes on with kids with special needs and if there's something on the telly you're looking and you're thinking oh that sounds like that oh I wonder what he's got and you're just trying to put a label on it yourself (Calum's father, 1)

Olivia's mother described being influenced by a programme on television when it came to thinking about her daughter's diagnosis and what might have caused her condition.

I mean they reassured me all the time I can hear them still saying to me you know the baby's fine the heartbeat's fine but all these programmes that I watch you know in America whenever a lady who is giving birth has Meconium staining they're very oh we have to get this you know very much get this baby out whereas Olivia was in there with this Meconium staining all this time and that must have been getting into her lungs she must have come out with a wee bit lack of oxygen (Olivia's mother, 1)

Following this, Olivia's mother requested a meeting with her consultant to go through her birth notes, although this was ruled out as a possible cause for her daughter's difficulties. Her understanding of what her daughter's diagnosis may be and what may have caused the disability is an iterative process that is influenced by information she accesses outside of the clinic, and interpretation and investigation of this information when she takes it back to the clinic.

A change in the parents' knowledge in the form of a new awareness could be the catalyst for search activity and the process of seeking diagnosis may be reinvigorated with new information that requires further work in and outside of the clinic.

If there's a change, either if you were to hear something in the news or something that you felt was linked then you might kind of go and look into it a bit more (Abdul's mother, 1)

Routine appointments were also reported by parents as being a trigger for increased search activity. Rupert's mother reported an increase in researching possible diagnoses before appointments.

Prior to any assessment coming up and prior to any meetings review meeting at the school or after letters that come in... I guess because I've got a review tomorrow I've been doing it again [researching online] and I'll be likely to flick around on websites now (Rupert's mother, 1)

More commonly, parents researched the information they had gained from the doctor or consultant following their child's attendance at a clinic or appointment.

Once we'd seen the genetics specialist and the paediatrician the first couple of times...we used to look up things when we came back from the meetings but now there's nothing we can look up to be honest (Calum's mother, 1)

If they said something specific in a clinic you know that we didn't recognise or something different it would probably trigger it off (Cameron's father, 1)

Parents were keen to research terminology they were unfamiliar with, signs or symptoms that were noticed or highlighted in the appointment, and any suggested or tentative diagnoses talked about in the consultation. In this way clinical consultations were triggers for resuming, maintaining or increasing searching activity.

We had an appointment just a routine appointment...they thought maybe there was a problem with one of the pupils responding and you know I'm instantly then on the internet trying to find you know any little glimmer of anything that anybody might say will set me off again looking (Eva's mother, IF, ENG)

If we have a consultation that's usually what would spark it cause you know...the consultants because they do sort of chat about stuff and after that [Abdul's father] especially will go and if they say something he'll look them all up and see what they are (Abdul's mother, 1)

A change in the health, development or behaviour of their child could also trigger increased search activity; 'Any new sign or symptom would start me off again' (Eva's mother, IF). This change could be a new sign or symptom or the onset of a new behaviour or issue.

Moy's general health or the onset of a new sign or behaviour will always get me started (Moy's father, IF, USA)

I start look for a diagnosis when Anna has issues that arise that I don't understand or there's an additional symptom to look up (Anna's mother, IF, USA)

Similarly, existing behaviours or health problems that were still an issue or that had recurred could be a trigger for increased searching.

One of the things that triggers us all the time is when she does this [name of behaviour] when she's quite clearly in pain with this stomach (Emily's father, 1)

Triggers identified that saw parents increase, maintaining or resuming their search activity were diagnoses suggested by others, routine appointments, and any change in the health, development or the behaviour of their child.

Nearly all parents described how the quest for diagnosis had reduced over time. Although triggers that saw parents resuming or intensifying their search for diagnosis were described, implying that the process of diagnosis ebbed and flowed, most parents reported a reduction in their search activity over time. Three parents reported they had not reduced their search activity and were still as actively seeking to locate a diagnosis for their child. For one parent, her searching activity had increased over time. For a small minority of parents, therefore, the intensity of the search for diagnosis was maintained.

Parents reported various reasons for reducing their search for a diagnosis. Work, caring and other time pressures had resulted in less energy and time available for searching for a diagnosis for some parents. Sarah's mother, for example, said that being tired and exhausted and prioritising caring for her children and getting enough sleep over looking for a diagnosis had meant that she wasn't so 'passionately' looking for a diagnosis anymore. Abdul's mother, having just had her second child at the time of her second interview described how, in the run up to Christmas and with a new baby at home, time restrictions resulted in reduced searching for a diagnosis. For Moy's father, time restrictions caused by his work, alongside 'weariness' and 'frustration' could put a stop to his search for diagnosis; 'my work schedule will usually stop a search' (IF). There could thus be a strong emotional as well as practical aspect to the reduction in searching.

Just as negative changes in their child's health or development could trigger continued or increased searching for a diagnosis, so too could improvements in their child's health or development be a reason for reducing the intensity of the quest for diagnosis, as Emily's mother reported.

I mean her communication has come on dramatically so that helps, all these things actually help you know there is some kind of improvement there...so I suppose you're probably not as frantically looking for a diagnosis (Emily's mother, 2)

For some parents the urgency of the search for diagnosis was reduced if their child's health or development was stable, for example Abdul's mother said she didn't search when 'things



were in the status quo' (1) and Eva's mother related reduction in searching to times they were 'in a lull': 'I think when we're in a lull and everything's fine and she's going to school like all the other children ... then for me too it's sort of it's my calm time and it allows me to think about my work and other things [than diagnosis]' (TI, ENG).

The frustration of not getting anywhere with their search was also cited by parents as a reason for reducing their search activity. Emily's mother said 'I've probably gone as far as I can really go' and described the process of seeking diagnosis as 'like hitting a brick wall' (2). For her reaching that brick wall, 'the end of the line', meant having to reduce her search activity. She said that searching for a diagnosis had been 'wearing' and 'exhausting' and that this had been a factor in reducing search activity; 'its part of the reason why to some extent we kind of we backed off diagnosis'.

Peter's mother described a process of giving up hope that she would ever find a diagnosis, which impacted on her searching activity.

I have become less obsessed by it as I've kind of given up hope. So now I don't really spend much time on it (Peter's mother, IF, AUS)

She also highlighted the negative impact of the search activity, 'searching for a diagnosis is destructive', and therefore felt she 'should stop trying' (IF). Some parents felt there was 'nothing else we can look up' (Calum's mother, 2) or felt 'discouraged after a dead end' (Anna's mother, IF), having 'nowhere left to look' (David's mother, 2) and so their search activity decreased in response to their perceived inability to take the quest any further. They felt there were no more avenues that offered possibilities for reaching diagnosis, for the time being.

Parents reported that the negative emotional impact of searching for a diagnosis had contributed to their decision to reduce their search activity. Eva's mother, for example, felt that the search for a diagnosis had 'taken over'. As she wanted her daughter to have 'a happy normal childhood' (IF), she had much reduced her searching believing it was getting in the way of this. Olivia's parents spoke of reaching 'saturation point' (1) with searching for a diagnosis and decided it was time to stop pursuing diagnosis so intensively. The following quotes from both parents suggest that they actively decided to stop their search activity due

to the difficulties associated with searching and the impact the quest for diagnosis was having on them.

You could see that Olivia had a bit of this and Olivia had a bit of that so you're frantically, okay I'll look up this and I'll look up that, what does that mean then and I said I just sort of I can't do this anymore... so I just had to stop it (Olivia's mother, 1)

It got to a stage where we were just so tired with it that we couldn't actually go on, we made a decision that it was it was getting us down...we couldn't do it anymore without actually knowing, it was really draining....we kind of we were at saturation point and we had to draw a line under it (Olivia's father, 1)

I have suggested several triggers that saw parents increasing, maintaining or resuming their active search for diagnosis, as well as identified factors that contributed to parents reducing or stopping their active search. There seems to be an interesting tension between the diagnostic search as constructive and hopeful yet, after a time, destructive and hopeless. Considerable emotional and intellectual resources are needed for the search including time, risk of being upset or overwhelmed, access to the internet, and sorting through a large amount of evidence and information. Triggers that saw search activity resuming or increasing were often related to the need to make sense of things in a changing world. Change could come in the form of a new behaviour, symptom or health issue or a change in knowledge as suggestions and information came from both inside and outside of the clinic. When the status quo resumes, or a new status quo is found, so search activity may lull. In the next section I focus my attention more specifically on the genetic testing process, and consider how parents made sense of genetic testing, of possible genetic cause, and of the field of genetics as advancing.

#### **4.4 How parents made sense of the diagnosis process**

In this section, I look at how parents made sense of the genetic testing process. This focus of analysis is felt to be particularly important and timely, as genetic explanations are increasingly meaningful in explaining childhood disability and illness (McLaughlin and Clavering, 2011) and paediatric genetics is increasingly playing a role in explorations of

childhood development delay (McLaughlin and Clavering, 2012). All but three parents gave accounts of genetic testing, with 19 describing on-going contact with the geneticist. The three parents who had not experienced genetic testing were Tom's mother and Rupert's parents. As their experience was atypical, analysis in this section does not include data from the interviews with these three parents. Parents were not directly asked about their understanding of genetic testing. Themes reported here, therefore, arose directly from the data.

For all parents of children seen by the geneticist, it was reported that all of the tests or assessments available to their child had been carried out. Two parents were awaiting the results of a genetic test at the time of the first interview; in both circumstances this had come back as negative for the conditions being tested for by the second interview. This meant that for the majority of parents, living without a diagnosis was an experience characterised by an assumption that their child may have a genetic condition that it was not currently possible to test for. A parent's understanding of the testing process may be complex in that they simultaneously need to grasp that all available testing has been exhausted with no resulting diagnosis, and at the same time hold on to an awareness that things may change as new testing or knowledge becomes available.

#### *Parent understanding of genetic testing*

This section makes two key suggestions; firstly, that parents interpreted and understood genetic tests differently, with some parents seeing testing as conclusive or certain, and others as inconclusive or uncertain; secondly, that parents invoked different types of knowledge in the making of these assessments. In suggesting the latter, I refer to Santos' (2006) theory that people make use of syncretic knowledge to understand inherited disorder. His model links socially shared/everyday knowledge, or lay understandings, with scientific explanation. In doing so he modifies Piaget's (1959) concept of syncretism of explanation, suggesting the model of syncretic knowledge as a heuristic tool (Piaget, 1959 cited in Santos, 2006).

Parents had different ways of understanding genetic testing. Most parents perceived that a negative test result did not necessarily rule out a particular condition or genetic cause; however a small number of parents interpreted genetic test results as being conclusive in ruling out that their child had a genetic disorder. Three parents believed a genetic diagnosis had been ruled out and perceived genetic testing as able to conclusively determine whether or not the condition had a genetic aetiology. This view is seen in the narratives of David's mother, Cameron's mother and Ismail's mother; none of whom believed their child had a genetic condition following genetic testing. David's mother suggests the reason the geneticist considered genetic tests for her son was to rule out a genetic cause for his difficulties:

She was going to send him for some tests cause they wanted to rule out that it wasn't in his genes or anything the reason he's got learning difficulties (...) the test was negative...it's not genetic (David's mother, 2)

David's mother also refers to another source of knowledge, family history, which further augments her belief that the tests carried out rule out a genetic cause for her son's condition.

There's nothing in the family...all the blood tests and the Fragile X and all that kind of stuff it's kind of all kind of ruled out [.....] It's not a genetic thing I, I don't think he's got a genetic em you know disability...I mean there's nothing in my family (David's mother, 2)

Here, her understanding of how genetic conditions are caused are rooted in notions of inheritance and a belief in the ability of genetic tests to offer certainty about aetiology. Cameron's mother and Ismail's mother expressed similar views that genetic tests had ruled out that their son's condition was genetic: 'It's not anything genetic...because nothing showed up in his genes' (Cameron's mother, 1); 'He's been tested for genetics a lot...so it's nothing to do with that' (Ismail's mother, 1).

Several other parents were confused by the negative results of genetic tests, suggesting an expectation that the results of genetic testing should offer conclusive answers. Claire's mother, for example, said when asked why she thought her daughter didn't have a diagnosis:

I don't know because they've done all the chromosomes and everything's normal she's not got any extra or any more...they've counted her blood cells and everything....they've done everything they can possibly do (Claire's mother, 2)

She is confused by the lack of answers offered by genetic testing. It may have been that Claire's mother would have ruled out a genetic cause for her child's condition, if it wasn't for her family history. She describes how several of her family members have had difficulties, including difficulties in one of her own pregnancies that resulted in a condition that was incompatible with life. Yet all genetic tests have been negative. She suggests diagnosis may be possible in the future, when new equipment becomes available. So she keeps on the possibility that there is something, in her words 'doon the line', although has difficulty making sense of the normal test results.

it could be something me and Philip's it's one in every thousand that could meet up it could be genetic they just don't know cause they done all our chromosomes they done mine Philip's and Kelly's and ours were normal as well ... and I think they were going to look into the genes or something but there was no more equipment available to do any more testing (Claire's mother, 2)

Here, Claire's mother uses syncretic explanation (Santos, 2006) fusing scientific understandings with her own lay understandings to make sense of recessive inheritance, genetic testing, the advancing field of genetics, and the results of genetic investigations. This is illustrated in the way she explains these complex scientific concepts using 'everyday' language (Santos, 2006), potentially expressing what Wagner calls 'vernacular science' (2007, p7) knowledge. I suggest Wagner's concept of vernacular science knowledge can be extended so that it is relevant here. Wagner uses this term, amongst other things, to refer to groups of lay persons who have a stake in a particular technological issue that materially affects them, thus they develop expertise that is relevant to their problem, displaying an 'intermediate' level of knowing about science. By virtue of their experience, they become more expert. Claire's mother expresses a degree of scientific knowledge about genetics yet at the same time holds on to a lay understanding of genetic testing as conclusive.

Like Claire's mother, Olivia's mother also appears to perceive that genetic testing can offer conclusive evidence that everything is normal;

A whole lot of bloods were taken and they came back negative they had tested for a lot of different syndromes...and everything was grand you know there's nothing else wrong internally that they could find (Olivia's mother, 1)

Despite her expectations of genetic testing as able to show there is 'nothing wrong internally', she doesn't rule out a genetic cause for her daughter's disability as she has been given further information about newly developed additional tests that may be available subject to funding, and she has been told by the geneticist that some syndrome features don't become apparent until a child is older. This causes anxiety for Jessica's mother who fears her daughter will experience facial changes and other clinical symptoms. She finds it difficult to make sense of potential genetic aetiology due to her other child being unaffected and the lack of an 'answer' offered by genetic testing.

Why did it happen to Olivia? If it never happened on Olivia would it have happened to Emily? But then one doctor said to me, no if you had that same if you were to do Jessica's pregnancy all over again it would happen again you know but why? (Olivia's mother, 1)

As her understanding of genetic aetiology relies primarily on inheritance, she is confused about why this would happen to one of her children and not the other.

In contrast, around a third of parents who consulted with the geneticist specifically talked about the uncertain nature of genetic testing or appeared to have knowledge that genetic testing was not necessarily definitive. Abdul's parents, for example, both expressed awareness of the uncertainties and complexities of genetic testing. Abdul's mother recalled that the geneticist had shared with her that there were many unknowns in the field of genetics; 'The geneticist said you know we don't have all the answers and we think this is where we're at in our understanding' (1). She also described how, over time, the geneticist had changed his view on the chances of recurrence. Referring to the changing nature of genetic knowledge and the lack of certainty about the likelihood of recurrence, Abdul's mother said; 'they're learning things all the time and things change' (2). Abdul's father held a similar view and spoke of how, even in the time since his son was born only 4 years before, the knowledge of the geneticist had advanced; 'we had another follow up consultation with [name of geneticist] and he said, you know, in the obviously during Abdul's life their, you know, their understanding had moved on' (1). Abdul's father interprets this as

representative of the high amount of uncertainty present in the field of genetics and the significant gaps in genetic knowledge. Abdul's parents accepted that genetic test results were not conclusive and that knowledge about genetics was subject to change.

Calum's parents talked about the complexities of genetic testing and how some syndromes were difficult to test for; 'this is a very rare syndrome very very very hard to diagnose' (Calum's father, 1), 'we knew that it wasnae going to be 100% you know definitely' (Calum's mother, 1). Even though the molecular test had come back negative for the syndrome being tested for, they had not entirely ruled it out. His father acknowledged the complexities of the field and how an answer may be more likely in the future; 'genetics itself is a complicated subject...I think if we could travel forward 50 years they could diagnose him' (1).

By highlighting this theme of parent perceptions of certainty or uncertainty in genetic testing I suggest that parents made sense of the genetic testing process in different ways. A small number of parents believed a genetic cause had been ruled out following negative test results; others struggled to make sense of the testing process as they expected genetic testing to offer certainty or because their understandings of genetic aetiology were rooted in ideas about inheritance. Yet others, around a third of the parents who consulted with the geneticist, seemed to accept the uncertainties of genetic testing and the inability in many cases for genetic testing to offer conclusive answers.

Parents drew on different kinds of knowledge when trying to make sense of the testing process. Prior lay beliefs that compete with current scientific knowledge can effect an individual's grasp of genetic concepts (Driessnack et al., 2014). In this era, described as the intersection of the genomic era and the information age (Driessnack et al., 2014), it is even more possible to fuse different kinds of knowledge in the effort to make sense of a medical experience. For some parents, as described above, fusing different kinds of knowledge led to difficulties making sense of genetic testing as their prior beliefs did not synchronise well with the scientific explanations they encountered.

Here, I look more closely at how parents made sense of genetic causation. I suggest that parents had different ways of making sense of genetic attribution; with some parents referring solely to explanations of heredity, and others going beyond explanations of heredity in referring to non-heritable genetic disorders. As with understanding genetic testing more generally, parents made use of syncretic explanation to understand genetic aetiology.

With reference to understanding genetic causation within the framework of heredity, a small number of parents (four) referred specifically to carrying a defective gene or the genetic condition being something they may have 'had' or carried. Claire's mother wondered whether it was something 'me and [partner's name]'s got' and believed there may be 'something doon the line'. For Emily's father, who had a relative with Fragile X syndrome, a negative test result for Fragile X for Emily did not rule out the condition in his mind. For him, the family history was too significant to discount when considering Emily's diagnosis and his belief in genetic conditions as heritable carried more significance for him than a negative molecular test.

A small number of parents relied on explanations of inheritance when accounting for potential genetic aetiology. Around a third of parents (8), however, had understandings that went beyond explanations of heredity. These parents made sense of genetic attribution with reference to a range of abnormalities of the structure or function of genes or chromosomes, bringing their own lay interpretation to often complex scientific concepts including chromosome abnormalities like numerical disorders, structural abnormalities like deletions, duplications, insertions and translocations, as well as single gene disorders.

Andrew and Lee's mother, for example, considered genetic diagnosis in light of new available testing and made reference to the structural abnormality of chromosome deletion. Although the test was negative, she displayed knowledge of non-heritable genetic disorder.



There was a new blood test came out to show out to show up any chromosomal you know big bits missing little bits missing no there's nothing like that (Andrew and Lee's mother, 1)

Here, and elsewhere in her narrative, Andrew and Lee's mother describes the genetic testing process using both scientific ('autosomal recessive disorder') and lay/everyday understandings ('big bits missing'). This kind of syncretic explanation is seen in other parent narratives. Olivia's parents, in trying to make sense of the information the geneticist has given them, also use both scientific and lay/everyday explanations as they struggle to fully grasp what may have caused their daughter's disability. Both refer to gene mutations and talk about deletion and duplication of genes and chromosomes.

[referring to a test carried out for Angelman's syndrome] it can be traced back to the mother being missing certain chromosomes or the father producing two chromosomes (Olivia's father, 1)

it could be a rare gene problem that's missing she could have a gene that's missing from this Angelman's syndrome (Olivia's mother, 1)

Abdul's mother and father describe in detail how they make sense of the information given to them by the geneticist. They report that they are initially told by the geneticist that the chances of it being genetic are 'probably quite small' (Olivia's mother, 1), as they are from very different geographical areas (Pakistan and Scotland), an explanation of genetic causation that relies on heredity. However around two years later, when they revisit the geneticist keen to have another child, this information is revised and it is reported that the geneticist has new knowledge.

And then eventually they said we think it probably is some sort of genetic thing (....) They thought the type of problem it was it might have been inherited from us .... but then we had a follow up appointment with [regional geneticist] and he said...during Abdul's life their you know their understanding had moved on and em that actually they're learning more and it might not be to do with a specific gene but more as how the g how the I don't understand but he sort of sometimes they credit you with a lot more understanding than you have but [laughs] it's more to do with how you know it's more to do with how you know it might have been a problem when it was formed em in which case it could be as likely to happen to someone else (Abdul's mother, 1)

Contrary to their understanding in the first year of Abdul's life, the possible cause of the condition is now understood to be something that was not necessarily heritable but a problem that occurred when the gene was formed. Information given by the geneticist is supplemented with reading outside of the clinic. They construct their understanding of genetic attribution using syncretic knowledge; scientific knowledge is fused with lay/everyday understandings so that a complex idea can make sense.

I think it's something to do with the fact that they don't think it was an actual defective gene but more in how it's been joined or something there's been some em thing that's affected a range of genes (Abdul's mother, 2)

He was saying that it's you know something has happened at some point ...he said it could have happened at any point he said it could have happened either to the egg or the sperm or or even as a fu as a fused thing so I mean and again I've I've I kind of look at books occasionally but there's no way I'm going to be able to absorb a genetics text book em it's I read sort of popular science but that's as far as it goes [laughs] I understand one thing he said to me ... it's to do with how eh DNA bits of DNA swap between things cause it's not just a combination of your male and female cells but it's actually bits of fragments of DNA can swap between the male and female parts apparently and I remember reading a book and I understood it at the time but it's gone... it's not as simple as A plus B it's kind of there's bit of jiggling about that goes on apparently (Abdul's father, 1)

Here, Abdul's parents make sense of the concept of translocation, which they suggest has been discussed at their last genetics review with reference to the possible cause of the genetic abnormality. They translate the scientific concept of chromosome translocation into lay terminology so that they can make sense of the explanation, with Abdul's mother making sense of the mutation as occurring in 'how it's been joined or something' and Abdul's father as 'to do with how bit of DNA swap between things...there's a bit of jiggling about that goes on apparently'. Each qualifies their explanation by suggesting that they do not fully grasp the meaning of the genetic cause, with Abdul's mother suggesting the geneticist assumed her to have more knowledge than she did and leaving her explanation open to ambiguity ('or something') and Abdul's father referring to understanding the concept in the past but now this understanding 'is gone'.

In looking at parent understanding of genetic testing and how they make sense of possible genetic cause, I have suggested that perceptions differ between parents and also that parents make use of syncretic explanation to communicate their understandings of this knowledge.

A small number of parents perceived that genetic testing was conclusive and could rule out genetic causation, and relied on explanations of heredity to make sense of genetic cause; a larger number of parents (around a third) were aware of the uncertainties of genetic testing and had understandings of genetic attribution that went beyond explanations of heredity.

#### *Parent perceptions of genetics as a changing field*

A significant number of parents suggested, or recounted suggestions made to them, that genetics was on the brink of significant change. In around half (9 of 19) of the narratives of parents who consulted with the geneticist, this theme of genetics as an advancing field was identified. The interview study was carried out in 2005-2006, prior to significant advances in genetic testing that took place in the years to follow. In parent narratives genetics is played as *becoming*, at an intersection in terms of being on the brink of significant advance that are not yet actualised. It is only since advances in technologies for genetic testing have come about in very recent years, with next generation sequencing technology and an ability to detect new genetic conditions through whole exome sequencing (Liu, 2010), that it is possible to put into context the advances that parents may have been alluding to. In particular, parents made reference to testing possibilities that were on the horizon but not yet available for clinical or commercial purpose. Soon after the interview study, exome sequencing to establish a diagnosis (including novel diagnoses) became available to families in the UK who fulfilled criteria for taking part in the Deciphering Developmental Delay research study.

Time-based restrictions on finding a genetic diagnosis were referred to by parents. That is, diagnosis was not possible now but may be in the future.

They have done every genetic test that they can do right now (Moy's father, TI, USA)

I think it's chromosomal problem they haven't got a test for yet (Caroline's mother, 1)

It would appear to be a genetic problem but science isn't there to do it yet (Andrew and Lee's mother, 1)

Restrictions on being able to diagnose their child were temporally related, as genetic testing and knowledge was not able to locate a diagnosis *yet*. At times the parent appears to directly paraphrase the geneticist in referring to the time-based nature of current genetic knowledge and testing available.

He [geneticist] said we don't have any way of testing *at the moment* you know (Abdul's father, 1)

He [geneticist] said but I'm almost certain that I'm not not going to find anything from tests that we can look at at the moment (Eva's mother, TI, ENG)

A number of parents specifically referred to 'equipment' becoming available to carry out testing. Claire's mother, for example, made reference to testing to 'look into the genes', possibly alluding to genome sequencing, but equipment was not available to carry out this testing; 'I think they were going to look into the genes or something but there was no more equipment available to do any more testing' (2)

Eva's mother recalled that the geneticist had suggested bringing Eva back to the clinic when she was older; 'to see if technology has moved on enough to provide any new tests' (IF). Abdul's father talked most explicitly and in most detail about the advances expected in genetics that may allow for improved genetic testing and therefore diagnosis. In our first interview, Abdul's father directly referred to gene sequencing. He was recalling information he had been given by the geneticist.

We saw the geneticist ... two weeks ago...and he was talking about he'd talked about em doing some gene sequencing thing i think em but unfortunately the equipment still isn't up to scratch (Abdul's father, 1)

The future of genetic testing and improvements in the process of diagnosis is tied to the improvement and acquiring of the equipment.

He was saying that the problem they have at the moment is that they they don't have the equipment they haven't got the equipment sorted out in terms of being able to do the thing reliably by the sound of it (Abdul's father, 1)

Abdul's father refers to 'spreading out your diagnosis area wider' (1) by improvements in equipment allowing for gene sequencing. He has supplemented the information the geneticist has given to him with his own reading, gathering and connecting knowledge from different sources in order to make sense of the genetic testing process and work going on to improve equipment so that testing is more time efficient. Furthermore, he also understands that while such techniques may offer the possibility of diagnosis, i.e. knowledge of where a genetic anomaly is located, this would not necessarily lead to increased knowledge about attribution or other information.

It's fairly clear that the tests and equipment will become available over time that can sequence key parts of his genes and look for those anomalies but again I mean that's probably a good few years away ...I think it might be decades away before we can like you know I was reading New Scientist some guy's come up with this scheme apparently em that would help to do sequencing quicker and stuff and clearly there has to be a reason why you'd want to do it and I guess the equipment would become more widely available over time but again that would just show you where the flaws are it may not tell you why and a whole load of other things again (Abdul's father, 1)

In our second interview, six months after the first, the family had again consulted with the geneticist and Abdul's father reports that the equipment for new testing is now available. He is, however, unclear about the time frame for testing and what kind of genetic mutation is being tested for, therefore does not fully make sense of this new process of genetic testing.

He [geneticist] said they've got the equipment now or something they said it was underway or in progress so I guess at some point it'll happen (...) I'm not even sure if I'm correct I'm probably making this up but I think they're looking for certain things in these things or certain sequences of patterns or whatever and I guess until they have that full thing yeah I would still think I can't imagine that you know it's going to be like you put something in a machine and out pops an answer I don't think that's anywhere near yet (Abdul's father, 2)

Abdul's mother expresses similar awareness of new testing techniques available, and confusion about the exact nature of the new tests and the time frame for carrying them out.

In around half of the narratives of parents who consulted with the geneticist, a dialogue of advancing knowledge in the field of genetics was apparent. Parent perceptions of genetics as a changing field appeared to be driven by the information given in the genetics clinic, with

some parents supplementing this with their own reading. New tests and equipment were referred to, and a commitment to a future in which diagnosis may be possible was suggested. In this way the process of diagnosis is open-ended; to be resumed. This notion of diagnosis as deferred is considered again in Chapter 6, looking at how parents made sense of living without a diagnosis. The deferral of diagnosis emerged as a key feature of the experience of living without a diagnosis for parents of children who were suspected to have genetic disorders. This playing of diagnosis as deferred rather than absent meant that the process of diagnosis had not an end, but a future.

## **4.5 Conclusion**

In this chapter I have considered the parents' active quest for diagnosis. In doing so, I have described different ways in which parents engaged actively and iteratively in the process of diagnostic searching, in seeking out and interpreting information and knowledge relating to diagnoses and in taking the results of their search activity back to the clinic. With the internet as the primary tool for search activity, parents joined in the detective work of diagnosis, seeking a label that was precise and definitive and that would help them to make sense of their child's difficulties. An emotional element to their engagement in the process of diagnosis was described, and one of the risks of searching extensively for a diagnosis was that they became overwhelmed, confused or upset by the information they encountered. Although much of their search activity was carried out alone, a feature of their quest was also the seeking of others to help them on their journey to find diagnosis. Times of change, for their child and in their own knowledge, were the catalyst to increased search activity. Different factors contributed to reducing or stopping the search, including practical considerations like time restraints and emotional responses to the search process. A sense of hopelessness was felt to characterise decisions to stop searching for some parents. Commonly, the quest for diagnosis reduced over time, although it ebbed and flowed and remained open-ended. There were differences in how parents made sense of the genetic testing process in relation to perceptions about the certainty or uncertainty of genetic tests and understandings of how the genetic disorder may have been caused. Parents made use of syncretism of explanation to understand this genetic testing process. A significant number of

parents perceived genetics as a changing field and appeared committed to a belief in the progressive nature of genetic knowledge.

Many families with a disabled child embark on a major quest for a specific diagnosis (Watson, 2009). Consistent with other research with families without a diagnosis (Rosenthal, Biesecker and Biesecker, 2001; Lewis, Skirton and Jones, 2010), parents in my own study were highly motivated to seek and obtain diagnosis and most (20) described an intensive search for diagnosis. This intensity was often more marked in the early years of their child's life or soon after finding out their child had difficulties. Parents engaged in extensive information work and, in doing so, were able to contribute to the diagnosis process.

In using the internet to access knowledge that may lead to a diagnosis, parents utilised e-scaped medicine (Nettleton, 2004) and became active players in the process of diagnosis. Other authors report on the importance of the internet in enabling parents of disabled children to access information relevant to their child's health condition or caring for their child (Blackburn and Read, 2005; Gunderson, 2011). Parents of children with genetic disorders can use the internet as a resource to clarify and supplement what they learn from their doctors, as well as to open up a new world of involvement in the process and category of diagnosis they had not previously had access (Schaffer, Kuczynski and Skinner, 2008). The parents in my own study accessed information online as a significant part of their involvement in the diagnosis process; in a sense, shopping around for a diagnosis (Arskey and Sloper, 1999). Diagnosis can be a form of active interpretive work (Arksey and Sloper, 1999) that patients participate in and a process of proto-professionalisation (Shaw, 2002; Hilton and Slotnick, 2005) may occur in which parents become more skilled in making use of the medical terminology used by clinicians and in online resources so that they can better navigate the diagnosis process. In doing so, they learn to 'talk the talk of the clinic' (Latimer, 2007a, p113) and actively participate in assessing the validity of the diagnoses suggested in the clinic or encountered online.

Parents, in recalling their involvement in the process of diagnosis, described how they endeavoured to 'fit' their child into a particular syndrome or diagnostic label. They carried out the nosological work of diagnosis; sorting through information, trying to categorise and name. In clinical genetics, the work of categorising patients involves mapping patterns of

physical features in patients and their families and identifying clusters of associated features and symptoms (Latimer et al., 2006). The art of dysmorphology relies on reading the body for sign and symptoms, with pathology becoming visible through a process of alignment and exclusion (Latimer et al., 2006). Parents in my own study carried out some of this work of diagnosis; in a sense they practiced a kind of *lay dysmorphology*. In trying to solve the diagnosis puzzle, they sought to dismiss diagnoses that were a poor fit for their child's signs and symptoms, and kept on diagnoses that their child fitted well with. Their search for diagnosis involved a process of 'pattern recognition' (Hunter, 2002). Other authors suggest that parents of children under the care of the clinical geneticist can experience a remarkable level of parental participation in the clinical process, with parents researching suggested diagnoses and bringing the results of their searching back to the clinic (Latimer, 2007a).

Parents' interest in visual representations of other children in an effort to find a child who resembled their own reflects the visual culture of paediatric genetics (McLaughlin and Clavering, 2012; Shaw et al., 2003; Featherstone et al., 2005; Latimer et al., 2006; Latimer, 2013; McLaughlin, 2014). McLaughlin and Clavering (2012) look at how families examine dysmorphology images on the internet as an example of the visual process embedded in diagnosis that takes place outside of the formal clinic environment. They highlight that a range of websites exist that catalogue the features of different genetic syndromes and that families often make sense of genetic investigation by drawing on these online sources of information. McLaughlin (2013) reports that parents can attach profound significance to these online images and that examining the faces and bodies of other children can carry significant meaning for parents. The significance of seeking other children who resembled their own was evident in a number of parent narratives in my own research.

Parents reported difficulties with searching for a diagnosis online and suggested that they required additional input to make sense of the information they accessed. Diagnosis is a more complex act than merely fitting patients into prefixed diagnostic categories and clinical judgement is central to genetic diagnosis, with clinical geneticists skilled in the craft of *seeing* a syndrome (Featherstone et al., 2005) as they practice the 'art' of dysmorphology (Latimer et al., 2006). Given the rare nature of their child's condition, it is not unexpected that parents had difficulty locating a diagnosis. In addition to this intrinsic difficulty of diagnosing rare childhood disorders, the difficulties parents experienced also related to the limits of their



knowledge. Medical knowledge can be seen to consist of 'something in addition to the facts' (Millar and Wynne, 1988, p112) and the expert eye of the clinician, skilled in the 'craft expertise' (Millar and Wynne, 1988, p112) of making diagnosis, may be needed to help parents interpret information accessed online and ultimately decide the significance of that information to the quest for diagnosis. Prior argues that lay people are not experts and are rarely skilled in 'the business of diagnosis' (Prior, 2003, p45) and that experiential knowledge may be limited and idiosyncratic (Prior, 2003). Although parents had unique knowledge of their own child, and a wealth of experiential knowledge, without input from the geneticist they were potentially not able to fully understand or interpret information accessed online.

Parents were also potentially emotionally vulnerable to the impact of information accessed online. Parents researching their child's (genetic) condition online can come across information that they find alarming and misleading (Whitmarsh et al., 2007). Schaffer et al., (2008) reported that parents of children with rare genetic disorders may feel a sense of obligation to keep searching for information that may help their child, even though the impact of searching may be additional anxiety (Schaffer et al., 2008). Families may commonly conduct internet searches with incomplete (genetic) information and the information obtained from these searches may arouse anxiety until interpreted by a genetics expert (Reiff et al., 2012).

Diagnosis as a process changed over time for parents, with almost all parents describing a reduction in search activity over time, although the search for diagnosis also ebbed and flowed depending on a number of factors. It makes sense that a quest for diagnosis may not always remain at the forefront of parents' minds and a principle focus of their activity; indeed to continue normal life activities including caring for their child or children, a hiatus in the active search for diagnosis may be necessary. Cohen (1995) reports on triggers of heightened parental uncertainty in chronic, life-threatening childhood illness and identifies triggers of heightened parental uncertainty, including: routine medical appointments; body variability (changes in health or behaviour); keywords and provocative questions from others; and new developmental demands. Cohen suggests that for families with children with chronic illness; 'an actual crisis is not necessary to reactivate fears that always lie just beneath conscious awareness' (Cohen, 1995, p14). For the parents living without a diagnosis

in my own study, there were similar triggers for heightened awareness about diagnostic uncertainty.

As parents journey through the process of seeking diagnosis, they experience shifting perspectives with diagnosis moving between foreground and background depending on circumstances and triggers. From a phenomenological perspective, this may be akin to awareness of the body in illness. When we experience illness our body moves from the background to the foreground of our awareness and our intentionality is directed to our body-self (Heidegger, 1962). When our body is functioning in the way we have become accustomed to or expect, our attention is taken away from the body and outwards (Carel, 2008). For parents of children with undiagnosed disability, when things appear stable for their child and in their knowledge, then thinking about unknown diagnosis may reduce and the quest to obtain a specific diagnosis may move into the background of their thinking and daily activity. Change may reinvigorate their awareness and interest in diagnosis and the quest begins again.

Parents made use of different kinds of explanations and fields of knowledge when talking about testing; using syncretism of explanation (Santos, 2006) to make sense of the genetic testing process. Parents also had different ways of understanding genetic testing and making sense of genetic attribution. Chapple et al. (2002), describing clients of the genetic counselling clinic as 'a lay audience', suggests that the technical vocabulary used by geneticists is often complex and confusing. Genetics can be an arcane subject for parents (Whitmarsh et al., 2007), obscure and mysterious. Even parents of children with genetic conditions who have spent a number of years carrying out their own research can still experience difficulties making sense of the 'genetics of the condition' which can remain abstract, with parents referring in general to mutations but expressing doubt about the details of the genetic information (Whitmarsh et al., 2007). Although parents in my own research referred to gene and chromosome abnormalities, their understandings in these areas were syncretic in that they relied on lay interpretation of scientific concepts, and they had some difficulties making sense of the diagnosis process.

Condit (2010) suggests that the public's understanding of scientific genetics overlap with professional understandings in some areas but not others, and that the public may rely on

the idea of genetic conditions being passed down through families; 'people understand genetics through the lens of heredity not in terms of the structural and functional nature of genes' (Condit, 2010, p1). In this way, there is a lay knowledge of inheritance that conflicts in a number of ways with scientific explanations, with lay understanding of genetics derived from concepts of the social relationships of kinship (Richards and Ponder, 1996). Most people have not had extensive education in genetics (Condit, 2010) and patients come to genetic counselling with varying degrees of genetic knowledge, a kind of 'lay genetics' (Armstrong, Michie and Marteau, 1998). Challenging these assertions, in my own research around a third of parents had understandings of genetic attributions that went beyond explanations of heredity. These parents made sense of genetic attribution with reference to a range of abnormalities of the structure or function of genes and chromosomes, bringing their own lay interpretation to often complex scientific concepts. This may be indicative of the prolonged nature of the process of diagnosis for parents, and their in-depth, on-going dialogue with the clinic.

A theme of genetics as a changing field was identified in around half of the parent narratives, with future genetic diagnosis relying on new tests or equipment becoming available following the expansion of knowledge. Since the interview study, significant technological advances have been made that may radically change the way developmental disorders are detected. Whole-exome sequencing was first reported in 2009 (Choi M et al., 2009); the results of this study demonstrated the clinical utility of whole-exome sequencing and had implications for disease gene discovery and clinical diagnosis. Further studies reported success in locating diagnosis using these new genetic testing techniques, for example a study in 2010 (Sarah B Ng et al., 2010) reported the first successful application of exome sequencing to discover the gene for a rare mendelian disorder of unknown cause, while a study the following year (Worthey et al., 2011) reports the power of exome sequencing to establish a definitive diagnosis in an individual patient in the setting of a novel disease after all standard diagnoses were exhausted (Worthey et al., 2011). It is now possible in the UK to carry out whole exome sequencing for unidentified childhood disorder in a clinical research setting. At the time of writing (October 2014) a study is underway in the UK involving 10,000 UK based families, the purpose of which is to 'advance clinical genetic practice for children with developmental disorders by the systematic application of the latest microarray and sequencing methods' ([www.ddduk.org](http://www.ddduk.org)). As of October 2014, nearly a third

of the first 1000 'trios', mothers, fathers and child, had received a diagnosis (Deciphering Developmental Disorders Third Annual Family Newsletter, October 2014).

The changes in genetic testing techniques alluded to by the parents may now be apparent. New genetic technologies allow a new level of variation to be seen and possible diagnoses to be made which were not possible before (McLaughlin, 2014) and improved detection of genomic alterations is made possible by increased sensitivity of chromosomal microarray (CMA) technology as compared to previous diagnostic technologies (Reiff et al., 2012). Other authors report that parents of children with suspected genetic disorders are committed to future scientific advancement and a belief in improved testing (Reiff et al., 2012). This is consistent with attitudes expressed by a significant number of parents in my own research. It is only now, in retrospectively considering how some of the parents made sense of genetics as a changing field, that we can contextualise their optimistic forecast about genetic diagnosis as accurately predictive.

In this chapter I have explored aspects of the process of diagnosis for parents. In referring to process I have kept in mind the multiplex dimensions of process: as a series of actions or steps taken in order to achieve a particular end, as a series of changes, and as influenced by the passing of time. In being agentive in the search for diagnosis, parents took action in order to achieve diagnosis for their child. They worked, often intensively, towards the telos of diagnosis as the imagined end of their diagnosis journeys. The search for diagnosis ebbed and flowed, and commonly reduced over time. However, as a process it did not end but changed. Over time, perhaps the quest became a kind of status quo. The journey, an open-ended diagnostic odyssey, may practically and psychologically not be sustainable at the level of intensity it begins with. While the intensity of the search for most parents reduced over time, they potentially remained vigilant for new diagnostic opportunities. In the next chapter, chapter five, I will explore parents' narratives. In looking at their stories of non-diagnosis, I suggest a core narrative of unresolved quest and identify a journey metaphor of stasis.



**5.1 Introduction: Why narrative research?**

**5.2 The kind of stories parents told me**

*The structure of stories*

*Restitution and the new normal*

*Unresolved Quest*

*Times of Chaos*

**5.3 Metaphor**

*The journey metaphor of stasis*

**5.4 Conclusion**

## 5.1 Introduction: Why narrative research?

In chapter 4 the parents' quest for diagnosis was examined and I considered how parents were agentive in the search for diagnosis and how they made sense of that process of diagnosis. Before moving on to examine data relating to the perceived consequences of absent diagnosis, the parents' narratives of non-diagnosis will first be explored. For all other data analysis, I carry out thematic analysis, parsing parent narratives into nodes and identifying themes in and across narratives. This thematic analysis is held to be crucial for organising the raw data generated by the research and enabling rich description of the participants' feelings, perceptions and experiences so that they may be shared with and understood by others. However, it is recognised that thematic analysis also fractures the story told by the parent in the interview. Arthur Frank (2010) recalls why he journeyed from carrying out thematic to narrative analysis. Speaking about carrying out thematic analysis early on in his career as a researcher he says;

The storytelling got lost as sound bites were extracted from different stories. The results were not false in the sense of being untrue; people had said those things about their illness...but it was unfaithful not necessarily to the content of the stories but to the art of the storyteller (Frank, 2010)

In this chapter, I consider the whole story of the individual parent participants by examining parents' narratives and the metaphors used in their narratives. In doing so, I suggest that understanding of the lived experience of the parents is enhanced. In carrying out narrative analysis the interview is taken as a whole and set in the context in which it has been generated and told (Bury, 2001). In preparing for this chapter, I considered each parent's story as a whole. Reissman (1993) suggests it is important to preserve not fracture narratives in research; 'because [stories] are essentially meaning making structures, narratives must be preserved, not fractured, by investigators' (Reissman, 1993, p4). While I embrace a narrative approach here, unlike Frank (1995, 2010), I do not forgo thematic analysis. Both approaches are seen to complement each other; with thematic analysis seen to be crucial in making sense of themes across accounts and narrative analysis adding depth to the understandings gained by thematic analysis and in maintaining the integrity of the *whole* story of individual parents. Research participants often turn to stories when asked to describe disruptive events such as illness (Stephens, 2011) and stories told by participants in research can convey the heart of

what is being studied. A more complete analysis of lived experience is possible by considering not just the *whats* but also the *hows* of storytelling (Holstein and Gubrium, 1995), and by letting the stories of the participants *breathe* (Frank, 2010).

The increased focus on narrative approaches in social science, seen in the last three decades in particular, has been described as a 'narrative turn' (Czarniawska, 2004). Accessory to the long literary tradition of studying the art of narrative, is the growing interest in narrative in the social sciences. This narrative turn in the social sciences lacks homogeneity of heritage (Hyvarinen, 2006), although Mishler's (1986) influential text on research interviewing is credited with bringing narrative to the core of collecting and evaluating social research (Hyvarinen, 2006). This relatively recent fascination with narrative in social science research gained momentum in the 1990s. The ensuing contemporary awareness of the significance of narrative in qualitative research extends across divergent areas and academic disciplines and so we come to be living in *the postmodern narrative's moment* (Maines, 1993) in the social sciences. There is also a growing popularity of narrative analysis in sociological studies of illness, and the emergence of 'narrative medicine' (Charon, 2006). The place of illness narratives (Kleinman, 1988) in the work of medical sociology was well established by the mid-1990s. A significant body of work, both inside and outside of social science, suggests the importance and ubiquity of stories and narrative.

We are the storytelling species. Storytelling is in our blood. We think in story form, speak in story form, and bring meaning to our lives through story...we recognize now more than ever everyone has a story to tell about his or her life, and they are important stories (Atkinson, 2007, p224)

We all tell stories; the telling of stories is universal (Bury, 2001); most if not all societal activities could not take place without narrative (Gergen and Gergen, 1998). When we tell stories, we do so out of our need to create sense out of situations (Goffman, 1963). Alisdair MacIntyre (1981) describes humans as story telling animals in actions and practice and, according to Roland Barthes, 'there nowhere is nor has been a people without narrative' (Barthes, 1977, p65). The ubiquity of narrative is described by Barthes:

'Narrative is present in myth, legend, fable, tale, novella, epic, history, tragedy, drama, comedy, mime, painting...stained glass windows, cinema, comics, news items, conversation...narrative is present in every age, in every place, in every



society...narrative is international, transhistorical, transcultural: it is simply there, like life itself...(Barthes, 1977, p79)

Important stories are told when there is disruption; and when asked to describe disruptive experiences such as experiences of illness, people turn to stories (Stephens et al., 2011). According to Mattingly (1994), chronic illness can represent narrative loss and may fracture a person's life story. Bury (1982) describes chronic illness as a disruptive experience, and refers to the 'biographical disruption' (Bury, 1982) of the illness event;

Illness, and especially chronic illness, is precisely that kind of experience where the structures of everyday life and the forms of knowledge which underpin them are disrupted (Bury, 1982, p169).

The taken for granted assumptions and the explanatory systems used normally by people are fractured by the experience of chronic illness, and chronic illness represents a critical situation (Giddens, 1979). Giddens (1979) asserts that we can learn a great deal about day-to-day situations in routine settings by analysing circumstances in which those settings are radically disturbed. I suggest that like chronic illness, the experience of undiagnosed disability represents narrative loss; a biographical disruption. The theme of fracture will be further relevant when considering how parents made sense of living without a diagnosis in chapter six. Here, I refer to fracture in the context of biographical disruption. The structures of everyday life and the forms of knowledge underpinning them may be fractured for parents by the experience of absent diagnosis.

According to Bury, language and narrative not only help to maintain and create the fabric of everyday life, they are significant in the repair and restoring of meaning when lives are disrupted (Bury, 2001). Narratives are used to repair the disruption caused when culturally specific expectations about life are not met (Landsman, 2009, citing Becker, 1997). Narratives bring things together so that they make sense; 'narratives offer the opportunity to knit together the split ends of time' (Hyden, 1997, p53). In the face of adverse events, personal narratives are changed to maintain a comprehensible sense of self, which fits their new circumstances (Bury, 2001). Whitehead (2006) suggests that one of the factors that disrupts the reconstruction of narrative is diagnosis, and when individuals are unable to achieve narrative reconstruction identity issues remain unresolved. For mothers of disabled children, repair of disruption for may not necessarily be achieved by establishing consistency with

past experience, and may instead be stories of transformation (Landsman, 2009). With reference to how we grow up with narrative over time Frank, drawing on Bourdieu's concept of habitus, refers to 'narrative habitus' (Frank, 2010; also Frank 2007 interview p127), that is; 'the collection of stories in which a life is formed and that continues to shape lives' (Frank, 2010, p49). Narrative habitus is our inner library of stories, the stories that people 'grow up on' (Frank, 2010, p25). Such stories are templates for experience; and we 'hitch a ride' on them (Frank, 2010, p25). While people grow up being cast into stories, they may not be aware of them. With biographical disruption, these stories may become visible. As Giddens advises, sometimes it is only when something is fractured that it becomes visible (Giddens, 1979).

If stories are performative, bridging acts (Frank, 2010) that *do* something in the act of telling them, then it is important to pay attention to the kind of stories parents told. According to conventional understanding, something happens and then stories are told that represent those experiences. This is understanding stories as *mimetic*, in that they imitate life. But this mimesis can be seen as a reciprocal process, so rather than story imitating life instead life and story imitate each other ceaselessly and seamlessly (Frank, 2010). Narratives not only describe life but also shape it; an on-going and constitutive part of reality, rather than as a transparent window into people's lives (Gubrium and Holstein, 1997). A focus on parents' narratives of their diagnosis journey may add to the gathering accounts of illness and disability told through narrative and, in offering insights into the kinds of stories parents told me, I hope to honour their stories of undiagnosed disability. As researchers we are privileged to have participants' feelings, perspectives and experiences shared with us. Narrative analysis is an approach, not a panacea (Riessman, 2000), and in doing narrative research and analysis we should seek not to control the meaning of a story. We can never grasp a participants' whole story, for that would finalise them, and these stories are on-going.

Interpretation, in its hermeneutic and dialogical tradition, is less a matter of decoding stories than of seeing all the variations and possibilities inherent in the story. The narrative analyst opens him- or herself to these possibilities, in order to invite others to open themselves. Interpretation seeks not to say: all the story is here, analysed and stated in clear, explicit terms. Interpretation seeks not to stand over the story, speaking about it. Interpretation seeks to be an on-going dialogue with the story (Frank, 2010, p104).

The aim of this chapter is to interpret the stories of parents and see possibilities and variations beyond the key themes identified in thematic research; to open up some aspects of the stories that would not have been noticed had thematic analysis been the only interpretive act in this thesis. Examining the storied nature of the interviews is meaningful; stories are important in this thesis. In the interludes, I am self-reflexive about how I came to look for the stories of the other parents, with my own story the catalyst for this journey. Inevitably, the stories in this thesis are intertwined. The story of the researcher's journey, the stories of the parent participants and the story told about diagnosis in this thesis turn around each other, form self-contained circles of their own yet are connected; a coiled structure linked by bonds, a helix. Parents began their interviews responding to an invitation to tell their story in their own words. Each formed a narrative then, and told a story of themselves and their child, and their diagnosis journey. In their interviews, parents may have narrated their story for the first time, giving chronology and order to what may have previously been unspoken.

In this chapter I consider the diagnosis stories the parents chose to tell by drawing on Frank's (1995) narrative typologies and extending these typologies as the parents' narratives did not fit neatly into Frank's schema (1995). I will also explore metaphors in parent narratives. In doing so, depth of understanding of the individual journeys of parents is gained. In referring to parents' narratives or stories, I refer not just to the stories parents choose to tell me in the initial narrative section of the interview when they were asked to tell me their story, but also how they continued that story throughout the interview.

## 5.2 The kind of stories parents told me

### *The structure of stories*

Arthur Frank (2010) describes the structure of all narratives by drawing on American mystery novel writer and journalist Bruce DeSilva's succinct description of the structure of narratives:

Every narrative tale...has the same narrative structure...: A central character encounters a problem, struggles with it, and, in the end, overcomes it or is defeated by it or is changed in some way (Bruce DeSilva 'endings' cited in Frank 2010, p30)

The parents' diagnosis stories are no different in structure. They encounter the problem of the diagnosis puzzle, struggle with searching for its solution, and are commonly changed in some way by this experience. Kermode (1967) said that we tell stories with the sense of an ending. This sense of an ending was described by Mattingly (1998) as 'telos', with events within our stories belonging to an 'evolving movement towards a telos' (Mattingly, 1998, p84). This telos, Mattingly describes, may not be located at the literal end of the story; rather it surfaces through the narrative as a whole. The story is directed towards the outcome; 'where one wants to be...in reaching ones' desired object' (Mattingly, 1998, p85). The parents' desired object is diagnosis, their quest driven by the telos of finding a diagnosis.

Mattingly (1998, 2007) draws on the work of folklorist Vladimir Propp (1968/86) who tells us that a story is governed by a lack or a need which must be addressed, either caused by some sort of insufficiency or created in response to the actions of a villain who disturbs the peace (Mattingly, 2007 p417). The protagonist embarks on a quest in the desire to attain that which he lacks or needs (Mattingly, 1998). Frank describes how stories are told because something out of the ordinary happens that requires a response (Frank, 2010). The trouble of a story (Bruner 2002, cited in Frank, 2010) is seen to catalyst the action of any story, as the status quo is fractured.

Everybody agrees that a story begins with some breach in the expected state of things-Aristotle's peripeteia. Something goes awry...The story concerns efforts to

cope or come to terms with the breach and its consequences (Bruner 2002, p17 in Frank, 2010 p28)

In parents' stories, more than one villain disturbs the peace; the developmental difficulties of their child and being unable to locate a diagnosis both potentially breach the expected state of things. With absent diagnosis, the stories of parents are governed by a lack. To go in search of a significant missing object is a motif central to the quest story in literary narrative. The quest story is one of the oldest and most popular of all literary genres (Auden, 1961). The hero seeks an object, which has significance in different temporal domains, not merely the present. It is a precious, not trivial, object; 'to look for a lost collar button is not a true quest' (Auden, 1961, p40). In parents' stories 5 of the 6 essential elements Auden (1961) suggests characterises the quest story come into play: a precious object is to be found; it is a long journey to find it (or not) and its whereabouts is not known to the seekers; the precious object cannot be found by anybody, they must possess the right qualities; a test rules out those unworthy (or unable) of finding the object; and helpers are required who assist the hero in their quest, by virtue of their knowledge. Famous quest stories include *The Wizard of Oz*, in which the main characters each seek something meaningful; brains, heart and courage; and *The Lord of the Rings* in which the quest is for the One Ring. In quest stories, the hero often has to leave home to journey, and returns home after the missing object is found.

I suggest that parents' stories have strong resonance with the quest motif in a literary narrative. More relevant to the study of narratives in medical sociology, are Frank's typologies of illness narratives. Frank (1995) suggests that people tell unique stories, but that they compose these stories by adapting and combining narrative types that cultures makes available. He suggests three such narrative types: restitution, chaos and quest narratives, and that in any 'illness narrative' all three types will be told. Frank describes stories as a way of re-drawing maps and finding new destinations. A person with chronic illness may find that the 'destination and map' they had used to navigate before are no longer useful (Frank, 1995). Stories may repair the damage that illness has done to a person's sense of where she is in her life and where she may be going (Frank, 1995). Frank recommends maintaining hermeneutic openness when talking about stories using his typologies, considering them more a heuristic guide than procedural guidelines. The kind of dialogical narrative analysis he advocates is ambivalent towards interpretation. Frank (2010) suggests

interpretation can reduce stories rather than let them breathe and may finalise stories by speaking the last word about them. Narrative analysis needs to reject the mimetic project, the assumption that stories imitate reality independent of the stories, and instead seek to understand stories as 'authentic fabrications' (Frank, 2010, p89) rather than reports about a fixed reality. Heeding this advice, I have not forced interpretation of the parents' stories by trying to fit parent narratives into any one of the typologies suggested in his schema and I have adapted Frank's typologies to make them more relevant for the parent narratives. A dominant typology of an unresolved quest narrative is identified, however I also identify elements of the other typologies (restitution and chaos) and I have adapted each typology to reflect the narratives of parents. I suggest three typologies are identifiable in parent narratives: *restitution and the new normal*, *unresolved quest*, and *times of chaos*. I now consider each of these typologies.

#### *Restitution and the new normal*

The restitution story is a master narrative that is faithful to the modernist assumption or expectation that for every suffering there is remedy (Frank 1995). It is our culturally preferred narrative, behind which is medicine's single-minded telos of cure. Restitution means things are fixed. The form is familiar; we are sick, we are fixed, normality is restored. The end of the story sees a return to the status quo. The restitution narrative is one in which 'modern medicine is the subject and the star' (Weingarten, 2001). Restitution narratives are reflective of Western medicine's emphasis on diagnosis, treatment and cure. Using the biblical story of Job as an example, Frank (1995) highlights that in such restitution endings the nature of suffering changes from a mystery to a puzzle. A mystery needs to be faced up to, a puzzle can be solved. However, when impairment is chronic, restitution stories no longer work.

In many ways parent narratives lay outside of the domain of restitution. There was no return to the status quo, the state prior to the 'illness experience'. None of the parent narratives were dominated by restitution, however it was possible to recognise elements of restitution stories which may reflect the desirability of this narrative type. This emerged in two ways; at boundary moments in parents' narratives in which a return to the status quo was still

achievable and desired, and beyond this to a form of restitution I have called 'the new normal' as parents celebrated their child's developmental achievements within readjusted parameters of normality.

In the first circumstance, there was temporal significance to when parents were most likely to express ideas of restitution. Those parents who described an early period in their child's life in which it was not known with certainty whether their child had a significant disability or not, expressed ideas of restitution in the form of being reassured by themselves or others that their child's development was or would be in the future within the acceptable boundaries of normality. In these stories, had this been the case, 'normality' would have been restored (or seen to not have been disrupted in the first place) and the concern they had for their child would have been alleviated with time passing and their child's development improving. These hints at restitution in the face of developmental difficulties are heard at times when the parent narratives are at a fault line. All of the narratives (other than Tom's mother and Rupert's parents for whom it was not relevant) moved beyond this fault line and significant impairment is confirmed. For many parents, their journey involved moving through this period when restitution was still possible. More than half of the parents described being reassured by others, most commonly health care workers, that their child's development was normal or could become normal.

I distinctly remember him saying that Jessica seemed to be fine and that with a few sessions of physiotherapy she would be fine. As it turns out he couldn't be more wrong (Jessica's mother, IF, AUS)

Several parents were reflective on the part they may have had to play in believing that a return to a state of normality would occur as their child's difficulties improved or did not become more significant. Cameron's father, for example, described how they were 'gullible parents' and so believed the reassurance others offered. They initially accepted that Cameron was slower to achieve milestones because of his gender.

He seemed to be quite delayed in other things compared to other children that we knew so I mean although although we were aware of these it wasn't a concern at the time because we were told different things, like boys are sometimes slower than girls and you know all these things and being, I suppose gullible first parents eh, we took we took all this in you know (Cameron's father, 1)

David's mother recalled that she was 'trying to put off' following up the difficulties she noticed her son had. It is her son's nursery who eventually suggests referral to the health visitor, as she continued to believe he might 'catch up' with his development even though she felt his behaviour to be different from other children. So while she *knew*, she hoped things would change and his behaviour would normalise.

He played on his own an awful lot and he was very repetitive in what he did em and the other thing I noticed about David was he he rocked a lot ... I remember like people used to say to me oh that's something not right he should be doing that (...) and em he was giving very little eye contact and it wasn't til that the private nursery eventually said em we want to contact your health visitor about David and I was kind of like yeah I know I know and I was kind of trying to put it off and kind of say yeah well he might catch up he's doing really well he's really healthy (David's mother, 1)

Caroline's mother told how she allowed herself to initially believe her daughter didn't have significant difficulties, suggesting that losing her first born child the year before Caroline's birth had an impact on her accepting Caroline may have difficulties. She delayed having her daughter referred to the occupational therapist following concerns about her development; 'we had a child before Caroline who em died aged four weeks from congenital heart disease so em...there was part of wanting em avoiding the inevitable if you like' (1).

For many parents there was an early period in which there were concerns but these were allayed by the reassurance of others or their own desire to maintain or return to a status quo of having a child with development within normal parameters. The complicating action of their narratives had occurred, but they continue to leave interpretation open to allow for the possibility that normality could be restored.

I also suggest that elements of restitution were heard in parent narratives in the way that a number of parent participants celebrated a development that they saw as allowing their child to live more normally. All parents were positive about their child's developments, but these parents in particular spoke of these new skills in the context of overcoming aspects of disability and achieving normative development, albeit to a lesser extent than their peers. I suggest this to be a narrative of *a new normal*. In the following 3 examples it is gross motor function that is celebrated. Having been told or believing their child may never walk or achieve other important milestones, these parents are celebratory of 'returning' to the



normality of having a child who can do these things. This also may relate to Frank's recent 4<sup>th</sup> narrative typology of *illness as normality* (Frank, 2010, p121). This plot involves minimising the illness, living in spite of the illness. This may be a common narrative for patients (or parents of patients) with genetic conditions who become expert at living with their genetic condition (Nowaczyk, 2012). However, I think the following examples go beyond this and it seems that the parents celebrate achieving normality of function that they worried their child would not have. In this way, restitution of sorts is achieved.

Olivia's mother, like many of the parents, spoke about noticing difficulties but not realising that something was significantly wrong at the time, which she explains may have been due to her being 'naïve' (1). A defining moment in her narrative is told as they get the results of a Magnetic Resonance Imaging scan (MRI) that shows an abnormality in her brain structure and they are told 'she will always be behind her peers' (1). Olivia's mother describes anxiety at this time about her development, as she isn't sitting up or crawling. As the story continues, it becomes clear that Olivia has made significant progress with her gross motor development. Olivia's mother describes balance problems and a wide gait, but also advances in her motor skills that see her being independently mobile when they worried she wouldn't be.

It's *excellent* she's came on *leaps and bounds* [her emphasis] and it's fab she's running and she's jumping (Olivia's mother, 1)

To an extent normality of development is restored albeit within the realms of a new normal; her daughter can run and jump, but with some balance and gait difficulties.

Andrew and Lee's mother also describes a moment of restitution when her daughter, whom they were told would never walk, takes her first steps.

When the consultant said to me take her home she won't walk she won't talk and I thought well I'm going to prove you wrong mate. So I did the best I possibly could and as I say we got her walking eventually she was ... maybe about three and a half first time she walked was across em ahh Trafalgar Square ... it was she just walked across looking at pigeons...that's the first time she got out of her buggy and we got her to walk without holding her hands or anything she just did it (...) That was a star moment that was, so we were able to walk into the back in to see the consultants and say chalk it on the wall we've done it (Lee and Andrew's mother, 1)

There is triumph as the goal they had been working towards, Lee walking, is achieved. Although her daughter continues to have significant disability in other areas, this aspect of normal development is achieved and can be 'chalk[ed] on the wall'. A walking frame is used for mobility after this, representing a new normal and celebrated; a negative prognosis is proved wrong. So while there is not a return to the state prior to the developmental difficulties, there is arriving at a place closer to it than had been anticipated.

Kamil's mother's narrative is particularly replete with references to milestones being achieved and these are celebrated as miracles. Kamil's mother chose not to have our interview audio-recorded. This made it difficult to incorporate detailed direct quotes from our interview into thematic analysis data chapters, which rely on the 'sound bites' of direct quotations to exemplify themes (Frank, 2010). In talking about Kamil's mother's narrative as a whole here, I am able to give more voice to the story she told me. Direct quotations are used but are limited in length (all quotes are face-to-face and from one interview). Over the course of our 5 hour interview she told me a story of a very difficult beginning, followed by a succession of miracles as her son achieved various milestones. Kamil's mother had a strong (Muslim) faith and made frequent references to God and God's will in her narrative. Like many of the other parents she initially described how she misinterpreted her son's early development; 'My baby could not do anything not turn his head or make eye contact to me it was normal. I thought this is what baby is'. She described staying indoors for 3 years, being very depressed, not talking to anyone and covering her son up if they had visitors through fear of them discovering his abnormality; 'they'd think he was like monster born not a human being'. A dialogue of aiming to maintain normality of appearance and function ran throughout her narrative. She decided against a feeding tube early in his life ('why should I make him abnormal?') and, following a period of physiotherapy, she noticed improvement in his development.

She began to talk of miracles when describing his development after his first year. His gross motor skills improved; 'he started to roly poly and at 2 sat up', which she described as being marvellous; 'it was like a miracle I don't have the words to describe it'. Another miracle she talked about was the development of his eating skills, describing an epiphany moment of him eating chapati following years of her liquidising his food; 'he took it, it was like a

miracle.' At age 3 he started to talk a little and further miracles are told: 'His nappy came off when he was 4', 'he's a miracle he talks on the phone.' She perceived the 'big numbers' and special dates to have symbolic significance in terms of him achieving milestones; 'at the big numbers something always happens, at 2, 3, 4. I'm waiting on 5. On Eid he walked for first time and stood up for a long time'.

A defining moment in her narrative is the birth of her second son, who does not have a disability. Following his birth, life becomes very different; 'life has just turned totally around'. Her story is one of restitution in this sense as she returns to a normality that she had missed out on with her eldest son; able once again to attend important social gatherings with her children. She tells me how 'it works now' when she visits people as her older son is taken by his father into the men's room and she can be in the women's room with her younger son. She has made a literal return to a culturally normal space, the room for the mothers and children. Another aspect of the restitution in her narrative is that in having a non-disabled sibling her eldest son appears less disabled, so he too is normalised; 'People don't think he's disabled. I dress them the same, why not?'

She has journeyed from a place of being excluded and isolated from the outside world ('When he was a baby, they talked about him') to being included and accepted in customary social events and practices and by her community. This is described as happening by virtue of Kamil's progress as he becomes increasingly more 'normal' ('look where it's ended for me it's a miracle ...now his nappy come off people think his brain is good... the outside world say look at him he's going to be normal') and by having a second son who does not have the disability. Her narrative is one of progress ('if there wasn't progress then I'd be crying out'), miracle, symbolic significance ('on 11th January Eid he stood up on his own for about half an hour he took his first two steps'), and belief in God's will and the power of prayer. Her belief in miracles defines her narrative of a restoration to a normality she missed out on after the birth of her first child and feared her disabled son would never achieve.

Other parents celebrate their child's progress and development, and express joy as they describe their child and family life, but for the parents described above the significance of their children achieving particular milestones seemed to be that something that may have been lost was restored. Not quite Frank's restitution narrative, but the achieving of a new

normal and thereby a kind of restitution. I suggest that in parent narratives it may be possible to identify aspects of restitution, even though a return to the status quo can never be achieved. This expression of restitution is heard in the voicing of hopes and dreams they had lost but still yearned for ('I have dreams of simple fishing trips with my kid' Moy's father, TI); in the dialogue of achieving normal development in the face of first noticing difficulties ('with a few sessions of physiotherapy she would be fine' Jessica's mother, IF); and in their telling how their child achieved a milestone that fulfilled normative expectations ('the outside world say look at him he's going to be normal', Kamil's mother).

Yet, this is only one reading of these moments in parent narratives. Equally, it is possible to say that a change in attitude may have occurred in parents over time that saw them experiencing self-transformation and therefore accepting a new kind of normal. I suggest that trying to categorise narratives using Frank's typologies is not straight-forward and the boundaries of each typology are fluid. Indeed, the typologies may be more flexible as an interpretation tool if they are seen to overlap. For example, the new normal theme identified above can be seen as both quest and restitution; a quest narrative in that the parents experience self-change as they accept a different kind of normal and can have optimism about this new boundary adjusted normality, yet a restitution narrative in that parents are celebrating the restoration of something that had been missing or that they thought may not be achieved in terms of normal development. To be meaningful, the typologies don't need to be distinct and it would seem to go against Frank's advice to maintain hermeneutic openness (Frank, 2010) by trying to force the narratives to fit any one typology. I suggest that considering the boundary moments in parent narratives in which restoration of normality may yet be possible, and the celebrations of a new normal, as restitution narratives may be helpful in understanding what was truly meaningful and yearned for by parents. Yet there is some cross-over with a quest narrative, in that self-transformation was experienced with regards to accepting a new kind of normal. Before looking more closely at the unresolved quest narrative form identified as the core narrative type in parent accounts, I first consider the *times of chaos* identified in a number of parent narratives.

Chaos narratives are the opposite of restitution: its plot imagines life never getting better (Frank, 1995). There is an absence of narrative order, coherent sequence or causality. The notion of remedy and progress are replaced by vulnerability and futility.

The story traces the edges of a wound that can only be told around. Words suggest its rawness, but that wound is so much of the body, its insults, agonies, and losses, that words necessarily fail (1995, p98)...Ultimately chaos is told in the silences that speech cannot penetrate or illuminate...Chaos is what can never be told; it is the hole in the telling (Frank, 1995, p101-102)

Telling a chaos narrative involves already having some distance and reflecting on an experience retrospectively in order to formulate it into a story. Therefore it may never entirely be a chaos narrative in that the telling of it requires a degree of retrospection; 'where life can be given narrative order, chaos is already at bay' (Frank, 1995, p105). In many ways true chaos narratives are anti-narratives, breaching the rules of chronology and causality. Chaos stories, therefore, cannot really be told although the voice of chaos can be identified.

Here, I have adapted Frank's typology of the chaos narrative (1995) and identify *times of chaos* in parent narratives. Moments of chaos were identifiable in all narratives, as parents recalled difficult times. However, for some parents more prolonged or intense times of chaos were evident in their narratives. Tom's mother, who had an experience of diagnosis atypical to the other parents' experiences, told a narrative dominated by chaos. Cameron and Jessica's mother told narratives in which elements of chaos stories were heard frequently or were particularly strong, and aspects of their chaos narrative were still present. All parents told of times that were difficult and expressed confusion, anxiety or sorrow about their child's difficulties as well as joy and happiness about their child's life. In all parent narratives it was possible to identify the voice of chaos intermittently, as you would expect with retrospective accounts of difficult times; in a very small number of narratives, however, this voice was a strong and regular feature.

One narrative that frequently told of times of chaos with reference to feelings about self-blame was that of Cameron's father. From the beginning of his story he made reference to

his painful feelings about how he may have caused Cameron's difficulties in the absence of any other explanation. Only seconds into his story he talks about his feelings of guilt and his narrative trails off into silence. His very first words 'without going into too much detail' indicate that there will be an unspoken element to his story of self-blame (all extracts interview 1).

Without going into too much detail I tend to get quite emotional about it because I blame myself a lot for it, and although I'm told by everybody that it's no my fault but it doesn't help the way I feel about it you know the way I feel that's it

NCD: And do you still feel like that?

Mhm

[silence]

However, he recovers his story quickly and elements of positive self-change can be identified that suggest some progress in coming to accept not having a diagnosis; 'it used to bother me more', 'the longer we go ... it's just sort of phased out the importance of trying to get a label'. When asked how he *felt* about not having a diagnosis he said; 'Years back I was very angry, at not having a diagnosis but now that it's been that long that we've it's still annoying now eh but I mean it's accepting it'. With reference to his own feelings of guilt about potentially causing Cameron's disability, however, there seems no movement forward and he shares that now he pretends to others that he doesn't feel as guilty but in reality those feelings are still as strong. At the end of his interview, when asked if he had anything he wanted to add he suggests that he has a lot more to say but does not feel it would be the 'right answers'; 'I don't think so no. I could obviously speak about this for hours you know but it's just having the right answers for the questions really so [silence]'. Further prompts are not successful and the interview ends. Perhaps Cameron's father's silence is the hole in the telling, the edges of a wound that can only be told around. While the chaos narrative is only seen surfacing at times in a narrative that is otherwise coherent, this element to this narrative is strong and temporally present and the silences associated with it suggest the depth of the wound. He seems stuck in the empty present of feeling to blame for his son's difficulties, which he describes has led to guilt and depression.

Times of chaos can also be identified in Jessica's mother's narrative, in which intense periods

of difficulty are recalled. She describes these difficult times having achieved some distance from the events and following recovery from depression associated with this time, therefore her narrative is not one only of chaos. Her daughter needed 24 hour care, screamed for hours at night, had several thousand seizures a day and would hurt herself badly on a daily basis. Jessica's mother describes this extremely difficult time, suggesting that they still have not come to terms with events (all of the quotes in this section are from our telephone interview, AUS).

It was probably yeah without question the worst time of my life and my husband and to this day we haven't dealt with it we live with the guilt and the sense of failure and and what happened and how is it that we were unable to look after our own child and you know what did we do wrong and how could we not do this

As Jessica's mother describes the severity of her difficulties she withdraws into a mode of chaos, finding it difficult to describe and breaking down emotionally in the interview.

I'd come into the bedroom and find blood dripping down the furniture and it's just I I I cannot even describe it now I *hate* the biting and the fact that she bites herself that that whatever is going on in her head is so bad that she would cause that kind of harm to herself ... but towards the end when she went away from home I always say went away from home but effectively we sent her away em it it just had got so bad and I just had to be with her all the time childcare weren't coping with her (...)it was awful awful awful and then she stopped eating and I think that just got to be the straw that broke the camel's back

At this point Jessica's mother describes how her ability to cope broke down and, with a need for 24 hour care and no support in place, she contemplated ending both her own life and her child's.

I was talking to my sister it was the day before Mother's day... and I was crying .. Mark and I were completely worn down and we were talking about how we would euthanise Jessica and I I know like I say euthanise but I I know it's murder we were talking about murdering our child we were talking about killing our child she was very very unwell we actually....because she was so sick and it was horrible it was an absolute nightmare ... every day was a nightmare we felt like we were living in hell and our world kept contracting and we felt like we were living in a prison ... I broke down I broke down at work ... I was talking to my sister and I was telling her what we were talking about doing about [sighs] euthanising Jessica and she said think about the consequences...I couldn't do it anymore ... I had no reserve left ... I broke down at work and luckily some people were there who were able to help

After seeing her GP that day the family travelled with Jessica to hospital, where she stayed for a long period of respite for her parents and after which she was cared for away from home with regular weekend visits home. The pain and guilt of leaving her daughter ('to leave your child with strangers') is clear and still strongly felt; 'that's how bad it was of me [very upset]'. This time of immense difficulty and pain is understandably extremely difficult for Jessica's mother to talk about and the chaos of the narrative in some ways lives on ('I cannot even describe it now'). Yet, the narrative only contains *times* of chaos, as Jessica's mother describes adjusting to living with her daughter being away from home. Even though her daughter's severe disability and need to live away from home is far from what she wants, she speaks of accepting that which she previously may have thought was unacceptable; 'I think I've learned to live with it, accept that it's unacceptable'. With reference to her having depression, a recovery story is described. Her recovery from depression is helped by supportive counsellors and medication. In the following extract from our interview she describes moving from a point of chaos ('I can't make sense of it. I feel like I'm going insane') to accepting the things that previously seemed irreconcilable.

When I got quite bad for a while I actually have seen counsellors and em obviously it was quite necessary and psychologists and was on anti-depressants for a time and the whole bit I remember saying ... I can't make sense of it. I feel like I'm going insane. There are these irreconcilable things in side of me you know where I love Jessica but I hate her or I hate what's happened because of her or what I've become because of her I I I don't know ... and she effectively said they are irreconcilable ... I think I've learned to accept that as best I can because I need to go on, I need to function em, I need to be part of my community and my society and my family and em, it's a bizarre kind of thing but I have accepted that's the way it is and I guess with the diagnosis it's the same. The wish for diagnosis I don't think will ever go away, human nature being what it is. I will always want to have and need to have that little box that I can put it in and tie a neat little ribbon on it and say yeah, that's what that is. I think I'll always have that but at the same time knowing that's highly unlikely I have to go on so I have to accept that is unlikely I'll always want it but it's unlikely that I'll ever have it

Jessica's mother's recovery from depression is a kind of restitution story. There is no return to the status quo, indeed quite the opposite with her young daughter now living away from home, but for her depression she describes a recovery and the restitution of mental health. She has also learned how to live with and accept her daughter's disability and, to some extent, absent diagnosis. She'll 'always want' but has had to accept living without diagnosis in order to 'go on' and be part of her family and community. This reluctant acceptance of



absent diagnosis over time will be more fully discussed in the next section on unresolved quest narrative. Here, I suggest that times of chaos were strong or frequent in only a small number of parent narratives, including Jessica's mother's narrative as she tells of devastating separation from her young daughter and accompanying feelings of sorrow and guilt.

Tom's mother opens her narrative by referring to a story type as she calls her son as a 'Jekyll and Hyde' character. The Jekyll and Hyde motif is embedded in Tom's mother's narrative from the beginning. A private clinic has recently diagnosed vitamin and mineral deficiencies and has suggested his diet causes the 'Jekyll and Hyde' behaviour. Tom's mother makes use of this story motif to define her son's behaviour. An assessment by the Child and Adolescent Mental Health Service is reported by Tom's mother to have come to a different conclusion about what caused her son's behaviour difficulties; 'they said em just a lot of em mental mental health in the family em ah and dysfunctional parent em [laughs a little]' (all quotes in this section are from one face-to-face interview). Tom's mother's narrative was more often a chaos narrative than any other parent, and she described no change over time in adjusting to or coming to terms with not having a diagnosis for her son's difficulties. This said, her experience is different to the other parents and her search for diagnosis is bound up with her desire to prove poor parenting is not to blame for his behaviour problems. Trying to prove her son has a medical condition, when specialists involved insist he doesn't, may contribute to her feeling stuck in the empty present of chaos. Her confusion and difficulty coping is evident, and she describes consulting with her GP following thoughts of suicide.

It's [not having a diagnosis] a big thing yeah cause I was always on the phone and asking em like coming round it was just I was just going round in circles and I was desperate and I was feeling like I I didn't want to be here anymore, cause it was really hard em (...) I went to see the GP this week ... cause I'm still not sleeping at nights very well, and he said that I had that I was suffering from exhaustion exhaustion and anxiety (...) he [Dr] wanted to refer me to a counsellor and that I just feel that [sighs] as time get goes on it's me all the time you know, that they're pointing the finger at me (...) it is it's it I feel worn down

She felt that not having a diagnosis meant that she was not able to prove that her son's behaviour difficulties were not caused by her own problems and parenting. Her narrative was of chaos and stasis; she was not able to move forward without a diagnosis and resolve the feelings of guilt and frustration. More than any other parent, Tom's mother seemed stuck

in an empty present with no promise that life would get better. When I asked her how often she thought about diagnosis, her reply echoed this chaos.

It's something that that dominates me [not having a diagnosis] I wouldn't say I'm I'm obsessed with it em but it's just something that controls me something I'm never never going to know an feel frustrated and I can't I don't know what I'm going to do an can go to college and stuff you know everything holds everything back em I never em know where I am with the family

For Cameron's father, Jessica's mother and Tom's mother times of chaos were frequent and intense. Although other parents recalled difficult times and shared journeys that had not been easy, the chaos narrative did not surface as intensively or frequently. Instead, the quest narrative was the dominant narrative in parent accounts. I adapted Frank's (1995) quest typology so that it was more relevant, and refer to *unresolved* quest as the core narrative.

#### *Unresolved quest*

Parents' narratives had strong resonance with the quest genre. Parents described a literal quest, the search for an answer – a unifying diagnosis. According to Frank (1995, 2010), quest stories are about change; they seek to use illness. They tell of being transformed; the teller has been given something by the experience, perhaps an insight. The final stage in a quest story is the return, when one is no longer ill but remains marked by the illness. In considering the quest typology, I explored whether parents suggested self-change as a result of their experience and what happened when restitution was not possible and the object of the search not found. There are three types of quest stories identified by Frank; manifesto, memoir, and automythology (Frank, 1995). The manifesto quest story claims prophetic insight, often carrying demand for social action. In the memoir, a gentle form of quest story, no special insight is claimed at the end, rather the illness is incorporated into the person's life. The third facet Frank calls the automythology, the predominant metaphor of which is the phoenix as the teller reinvents himself from the ashes of the fire of his own body. Individual change is emphasised, not social reform. Frank admits that the risk of quest stories is that they can present the transformation as too complete, and implicitly depreciate

those who fail to rise out of their own ashes. Frank utilises a phoenix metaphor to describe the change that someone undergoes during illness (Frank, 1993, 1995). Humans however, unlike the phoenix, cannot change completely. Those who experience little self-transformation are defined by Frank as reluctant phoenixes'. Smith and Sparkes (2005), writing about men and spinal cord injury, suggest that quest stories accept impairment and disability and seek to *use* it, the quest is defined by the person's belief that something is to be gained from the experience.

In the narratives of almost all parents (23 of 26) it was possible to identify self-transformation as parents, to varying degrees, came to terms with living without a diagnosis. Their quest was not resolved, diagnosis may be possible in the future, but search activity had reduced and most parents imparted a sense of learning to live with absent diagnosis. For some parents, the self-change described was little more than being less preoccupied with diagnosis or thinking less about diagnosis over time. The transformation for parents may have been reluctant, they would have preferred to have a diagnosis for their child but over time the urgency of their sense of needing a diagnosis had decreased. These are Frank's reluctant phoenixes (Frank, 1995). Their internal transformation may be at times unstable; their feelings about non-diagnosis possibly ambivalent. Two parents told a manifesto quest story, responding to their experience by setting up support groups for other families of children with undiagnosed conditions. Other parents' stories were memoirs, with little insight gained and more a sense of incorporating non-diagnosis into their lives, or were automythological, with individual change rather than social reform heard through their narratives. For the parents who experience individual change, like Frank's reluctant phoenixes, there was a change of character through suffering. Old 'intactness' was, as described by Frank (1995) stripped away to prepare for something new, with the rising out of the ashes coming from a move towards being more at peace with not having a diagnosis.

As described in chapter 4, the intensity of the search for diagnosis commonly reduced over time. Factors were identified that impacted on a parent's search for diagnosis, including time restrictions, emotional upset and the progress of their child's development. Here, I add to the analysis in chapter 4 by suggesting that some parents may also have reduced search activity because they adjusted to living without a diagnosis and were more able over time to

accept the unanswered questions of absent diagnosis than they had been at the beginning of their diagnosis journey.

I am accepting this more, that we may never get a unifying diagnosis (Sarah's mother, IF, AUS)

Parents' self-transformation, as they moved to accepting (to varying degrees) not having a diagnosis, had a temporal dimension with change over time described, as the following extracts from the interviews with Calum's mother illustrate:

As time goes by you just get used to a situation (...) you do kind of come to terms with it yourself so I think the longer it goes on the easier it'll be you know you will you will just kind of think oh well it's just one of these things (Calum's mother, 1)

It's just time isn't it it's like anything they say you know eh the longer you have to kind of accept something to come to terms with it the easier it becomes yeah (...) you get used to the situation ... a bit more well not completely but not completely but you definitely you do get used to it yeah (Calum's mother, 1)

Many parents used phrases that referred to how they initially experienced living without a diagnosis compared to how they felt at the time of the interviews: 'It was vital at first [to get a diagnosis (...)] now it's not the end of the world' (Calum's mother, 1); 'It was a bigger deal in the beginning' (Abdul's father, 1), 'It's not like a major thing now' (Claire's mother, 1), 'the longer we go ... it's just sort of phased out the importance of trying to get a label' (Cameron's father, 1). Most parents said they used to think about diagnosis more in the past, suggesting a move towards accepting living without a diagnosis and thus self-transformation over time. The degree to which this was apparent varied between parents. I now illustrate this variation in accounts of self-change by drawing on three of the parents' narratives. In these accounts, self-transformation ranges from the partial and reluctant change described by Jessica's mother, to the significant self-change told of by Calum's father.

In Jessica's mother's narrative self-transformation was discernible, albeit limited and reluctant. In the *times of chaos* typology section above, I told how she suggested her wish for diagnosis would never go away, but that she had to accept diagnosis was unlikely; 'I have to go on' (TI). Jessica's mother, however, was ambivalent about whether or not she had

accepted absent diagnosis. Elsewhere in her narrative she suggested she had adjusted to her daughter's disability, but not absent diagnosis.

We actually think we've adjusted really well to the disability' we've learned to accept though still sometimes be saddened by the fact that there are a lot of things that Jessica will never do. We love her anyway. But the lack of diagnosis is a bit like an open, never healing wound. And there is no answer to the age old why? (Jessica's mother, IF, AUS)

She realised she had to 'go on', and in many ways had done so, but making sense of things was still difficult without diagnosis. She still felt in limbo, suggesting self-transformation was partial.

So we're still in limbo and not having a diagnosis keeps you there you don't really you can't make sense of things (Jessica's mother, TI, AUS)

Emily's mother described how her attitude to diagnosis, like many other parents, had changed over time; 'the whole diagnosis thing it's there really in the background but eh it just isn't as dominant' (1). Diagnosis had moved from the foreground of her thoughts to the background. Triggers could see her thinking again about diagnosis, but the general trend was a decrease in significance of obtaining a diagnosis over time; 'you're probably not as *frantically* looking for a diagnosis it would be nice to have'. There is some ambivalence in her narrative about accepting absent diagnosis and along with describing adjusting to absent diagnosis she also describes being inspired not to give up. She makes reference to the film 'Lorenzo's Oil' which her GP had suggested she watch when she was going through a difficult time. Watching the film was said to have a significant impact; 'it held out that wee bit that said just don't give up you know you've got to be tenacious about this really' (1).

Emily's mother doesn't give up on diagnosis, but she gets 'off that train' (2) of needing a diagnosis so intensely. She keeps hold of diagnosis yet at the same time lets it go. Talking of both coping with her daughter's disability and adjusting to not having a diagnosis, she describes a change in mind-set. While still tenacious about helping her daughter, including discovering diagnosis, she has to let go of the ideas she used to have about family and things happening as you would expect them to. Emily's mother identifies that learning to accept

the life she has also has had a positive impact on her work and she can now empathise more with families she encounters who experience difficulties.

I mean people want to be able to have a kind of nicely wrapped up family and you know with everything that's hunky dory and family life just doesn't it doesn't work like that (Emily's mother, 2)

Calum's father spoke in detail about adjusting expectations of doctors and medical science. His self-change was very much related to becoming 'more realistic' about his expectations ('we've got a more realistic expectation now than eh than we had beforehand', 1). He described change over time in his attitude to not having a diagnosis; 'now we're not so hung up on it [having a diagnosis]....we have now adapted our own way of looking at it' (1). While diagnosis remains something that Calum's father hopes for, it seems to become less important to him over time. While having a diagnosis would be preferable, it's not as important to him as it was at the beginning of his diagnosis journey.

It would have been nice to be able to stick a label on him but it doesn't make any difference now to be honest it's just we know it's an overgrowth syndrome but fff it's just a case of we're now just hey ho (Calum's father, 2)

This is quite a change from the initial attitude to diagnosis he describes; 'initially we thought we were totally hung up on a label we need a label', 'it wasn't the be all and end all but it wasn't far off it' (1). The self-transformation he describes is a change to his personality and way of thinking about the world. At the beginning of his experience he said he was someone who 'like[d] to be able to organise everything...I need to be able to understand things to cope with them' (1). Absent diagnosis was said to 'go against the grain' (2) of this aspect of his personality, an aspect which has since been challenged and changed as a result of the experience of non-diagnosis.

Self-transformation was identified in most parent narratives, to varying degrees. A further dimension to this transformation as parents over time adjusted to absent diagnosis, was that it was often phasic. A general trend of decreased thinking about diagnosis over time was described, although around a third of parents also described a phasic element ('it goes in phases' Eva's mother, IF, ENG) to thinking about diagnosis;

We do go through spells ... where you know you'll kind of suddenly go oh! Right okay wait and I'll go and find out about this so [laughs] and at times I mean that wanes cause other things take over really and I suppose that's a bit less now (Emily's mother, 1)

It goes up and down depending on what's happening depends how Abdul is depends how you feel (Abdul's mother, 1)

For three parents (Tom's mother, Rupert's parents) the unresolved quest theme was not relevant and self-transformation in this context was not present in their narratives. For Tom's mother, who still sought to prove her son's difficulties had a medical cause, there was no evidence of change over time or of accepting a lack of diagnosis; for Rupert's parents this typology wasn't relevant as they were not seeking a medical diagnosis. For all other parents, a degree of self-transformation was suggested in their narratives.

Many parent participants described change over time; they told quest narratives. Most parents were Frank's reluctant phoenix; their preference was for diagnosis but they had to adjust to living without a diagnosis. Self-transformation was sometimes fragile, partial, and phasic. All parents for whom it was relevant were still seeking a diagnosis, to varying degrees. Thus the search for diagnosis, while potentially reduced over time, was never entirely called off. Almost all parents in this way told *unresolved* quest narratives in that they were quest narratives without closure. The object they left home to go in search of was not found, yet in some way they returned home with new insight. The following section takes this narrative analysis further by looking at metaphors in parent narratives. A core metaphor of *stasis* was identified in parent narratives. In this concluding section to the chapter I look at this core metaphor and what it might tell us about the parents' journeys.

### 5.3 Metaphors in parent narratives

Metaphor is an important way of using language to explain abstract ideas or to powerfully convey feeling and can be used as an interpretive tool to assist in understanding qualitative data (Cameron and Maslen, 2010). More than this, metaphors perform significant interactional work via narrative, crucial to the way people consolidate and extend ideas about their knowledge of the world (Becker, 1997). In this section I identify a core metaphor; the journey metaphor of stasis, and suggest that we can learn more about the parents' experience by considering this core metaphor.

Reddy (1979) suggests that the locus of metaphor is thought not language, and that metaphor is a major and indispensable way of conceptualising the world (Reddy 1979). Language is seen to function like a conduit enabling the transfer of thoughts, feelings, meanings, and ideas from one person to another. Building on this, Lakoff (1992) considers how metaphor involves understanding one domain of experience through a very different domain of experience, mapping the ontology of one with the other. Lakoff and Johnson (1980) suggest that metaphors structure experience, how we perceive and what we think and do, saying 'most of our ordinary conceptual system is metaphorical in nature' (Lakoff and Johnson, 1980, p4). Metaphor can be seen as an economical substitute for deeper knowledge (Tsoukas, 1991).

By investigating participants' use of metaphors, we can better understand their feelings, attitudes, and conceptualisations. Parents used an abundance of different metaphors in their narratives and when talking about living without a diagnosis and the search for diagnosis. As seen in other work on metaphors and illness (Sontag, 1978), a significant number of parents used military metaphors in their accounts. Ten parents made use of military metaphors when talking about trying to find a diagnosis. A 'push', 'fight', 'struggle' or 'battle' for diagnosis was described; 'we fought tooth and nail'. The search was said to be a 'minefield', 'a landmine', 'an uphill battle', 'there was a trap door waiting' with resulting lack of diagnosis a 'bombshell', 'like being punched in the stomach'. However, the most common kind of metaphor used by parents when describing their experience of seeking diagnosis, or living without a diagnosis, were metaphors of stasis in relation to movement. A core metaphor, the *journey metaphor of stasis*, was identified in parent accounts. I suggest that



this core metaphor of stasis indicated that parents search for diagnosis can be understood as a journey, with impediments to movement encountered on the way. The ontology of journey is mapped with the ontology of the process of seeking a diagnosis. Like a journey, they have a destination in mind. But travel is impeded, and stasis of movement results.

### *The journey metaphor of stasis*

Stasis in relation to movement was suggested powerfully in many narratives. These metaphors suggested lack of movement, of being stuck, of moving in unusual ways, of meeting obstacles impeding movement, and of difficult passage forward. Stasis was suggested through: metaphors of space including metaphors of conduits and perimeters; metaphors of containment, including metaphors of boundary and transition; and through metaphors of movement, including metaphors of direction, speed and acceleration and locus. Twenty parents used metaphors suggesting stasis in their narratives when describing the experience of living without a diagnosis.

Metaphors suggesting stasis, a lack of movement, included metaphors referring directly to being unable to move on or move forward. About a third of parents referred directly to being unable to move on or forward.

We are left no further forward (...) Getting a diagnosis would have helped us move on (...) We need to find out what's wrong with him because we could start moving (Calum's father, 1)

It doesn't really move on... I just want to say right this is what it is and let's move on (David's mother, 1)

[If I had a diagnosis] I could just move on a bit (Tom's mother, 1)

For some there was such a profound lack of movement forward that they suggested a sense of moving backwards in relation to diagnosis.

You get something in your mind and you think that's great and you prepare for it and then it changes and you think oh so I wouldn't think just treading water maybe two steps forward and three back (Calum's mother, 2)

I keep thinking we're going forward and we keep going back (Claire's mother, 2)

Several parents also used metaphors suggesting movement without getting anywhere. Around a quarter of parents talked of going 'in circles' ('I was just going round in circles' Tom's mother, 'it's just like a never ending circle' David's mother, 'we've done a huge circle' Emily's mother), with Calum's father comparing seeking a diagnosis to moving in 'ever decreasing circles'. Three parents referred to feeling as though they were in a state of 'limbo' ('at the moment we're kind of in limbo with her' Jessica's father; 'If you don't have a diagnosis you're almost like in limbo in a lot of ways you can't move on' Jessica's mother) with one parent suggesting living without a diagnosis was akin to living in 'no man's land'.

Metaphors of trying to move but being returned back to the place they started from were also used. Several parents talked of 'being back at square one' in relation to diagnosis and living without a diagnosis: 'We were pretty well back to square one' (Peter's mother). Cameron's mother, as described in chapter 4, used an analogy of making repeated attempts to climb a hill, only to be returned to her starting place at the bottom again as she failed to find a diagnosis. Metaphors of moving on difficult ground ('it's a rocky road' Moy's father, 'it's an uphill struggle' Cameron's father), or in difficult substances like water or mud ('it's like swimming in mud' Eva's mother) with little progress or help, were also used in parent narratives.

You're sort of just treading water for a while [long silence]. I think we'll always tread water, I think it's something we'll do for a very long time (Calum's mother, 2)

We're swimming about with nowhere to go I suppose with nobody throwing you a life line (Emily's mother, 2)

As well as difficult terrains, parents referred to obstacles preventing movement or causing stasis. Four parents talked of meeting or hitting their head off a brick wall ('we're still battering our head against a brick wall' Cameron's father). Other parents talked of movement forward being halted or distorted ('we're not getting anywhere' David's mother,

'we're being thrown in different directions' Olivia's father, 'you can only take so many knock backs' Calum's father), without identifying the obstacle causing stasis.

We were kind of stopped in our tracks...well you may never know [a diagnosis]...we may never ever get to know, and that kind em you know it kind of left you winded if you like (...) we were kind of cut down (Olivia's father, 2)

Emily's mother described not being able to locate a diagnosis as reaching 'the end of the line' using a variety of metaphors suggesting movement halted and a journey reaching an end. She knows she is not going to 'go any further' even though they have not reached their destination, diagnosis. Indeed, they are halted on their journey even though they don't arrive at a metaphorical station.

You feel as if you know after every kind of batch of tests that you know everything's normal so it's kind of the end of the line and I suppose that's the brick wall is the end of the line, you know your kind of train's arrived at the station and that's it, you're not going to go any further, or it is actually the end of the line rather than even a train station, it is actually the end of the line em [clears throat] cause you're not going anywhere so you have to jump the train (Emily's mother, 2)

Metaphors of darkness were also used by several parents when describing their search for diagnosis or living without a diagnosis: 'It's a journey in the dark...we're heading into a very very dark black hole... it's a bit like clutching in the dark just now' (Calum's father, 1). At times darkness acted as an obstacle potentially impeding successful movement: 'you're still swimming about really in the dark' (Emily's mother, 2)

Parents also made reference to doors, roads, avenues or paths. Sometimes doors were opened allowing for the possibility of movement forward or progress ('it's quite good that the door to genetics has been opened again' Emily's father, 2). More often, movement forward or progression on the journey to diagnosis was frustrated by being unable to travel down roads, avenues or paths.

There's no real diagnosis at at the moment or any one avenue that we can go down...I wasnae happy with em the fact that we were going a wee bit down this road and a wee bit down that road (Olivia's father, 1)

With a diagnosis you know exactly where you're going, your your future path is laid out for you and you can see that path relatively clearly and you can move on,

that's the important bit I think is being able to move on (interview 1) [...] It's like we're in the middle with no more doors to try, doors closing and there are no more to try. You come to the end of the road, it's a shut door. There was a chink of light with the [name of syndrome], but then the door gets shut again (Calum's father, 2)

The direction as well as the lack of movement was referred to by some parents, in particular being unclear about the direction to be taken: 'I thought there wasn't enough direction in what we were doing (...) we don't know where to turn' (Olivia's mother). The way forward was referred to as not being clearly marked or defined ('There are no guideposts' Emily's mother).

Several parents also used metaphors to suggest that their journey had no clear ending. Claire's mother, for example, described the experience of looking for a diagnosis and living without a diagnosis with reference to a rollercoaster, an unusual method of travelling a course that is circular, that was not going to stop: 'It's a rollercoaster, some pit stops along the way and that's it (2); 'It feels like a roller coaster that's never going to stop' (1). Jessica's mother spoke of their journey of looking for a diagnosis as being a route that had unusual direction and no end, describing how they 'embarked on a journey that has taken some unbelievable twists and turns and shows no sign of ending' (IF, AUS). David's mother felt the search for diagnosis was 'a never ending game' (1), while Emily's mother said 'if they cannot tell you what's wrong with your child then that's the end of the story' (1). Three parents also referred to the uncertainty of living without a diagnosis by using the spatial metaphor of not knowing what was around the corner.

A core metaphor of stasis of movement was identified in the majority of parent narratives. Parents used many different metaphors to suggest stasis of movement including metaphors suggesting lack of movement, being stuck, moving in unusual ways, of meeting obstacles and of difficult passage forward. I suggest that these metaphors indicate that parents conceptualised their experience as a journey, and further, a journey that was difficult to travel. The stasis metaphor may tell us that the journey was difficult, and parents felt 'stuck'; that there were many obstacles along the way that halted their progress. Indeed, when parents were asked how they felt about diagnosis in their interviews, many replied that they felt 'frustrated', one definition of which is the prevention of the progress or fulfilment of something. In using these metaphors the ontology of journey and stasis is mapped with the

ontology of the process of seeking a diagnosis and absent diagnosis. The ontological quality of a journey most relevant to parents may be the expectation that they will reach a destination. The diagnosis journey for them, however, does not reach a destination. And, like Emily's mother described, they may be forced to get off the train before reaching this destination, when they realise they are not going anywhere. Parents seeking diagnosis may feel the ground they travel on is precarious. They may travel in circles, finding themselves back to square one in a never ending game. They may journey in the dark, meeting brick walls, experiencing limbo, going one step forward and two back, trying to find open doors, treading water, climbing hills they never reach the top of, being thrown in different directions, swimming about with nowhere to go. If metaphor is shorthand for deeper knowledge, then from the metaphors used by parents we can know about the experience of living without a diagnosis that it was a journey; and further a journey on which movement was obstructed. This may suggest a strong underlying futility and vulnerability about the diagnosis journey, and explain the frustration that many parents experienced.

#### **5.4 Conclusion**

In this chapter, I have suggested the ubiquity of narrative and its important function. The parent participants in my research told stories of their child and their diagnosis journeys. As a compliment to examining the themes generated across narratives by carrying out thematic analysis in other chapters, in this chapter I have considered the *whole* story of the parent participants. By carrying out this analysis of narrative, I hope that I have shared aspects of their accounts that may have slipped through the net of thematic analysis and, in doing so, opened up aspects of their accounts that would not have been seen otherwise.

I have considered the relevance and utility of Frank's typologies (1995) and extended these typologies so that they were more relevant to the kinds of stories parents told me. I have also challenged Frank's typologies by suggesting that it is not easy or potentially desirable to consider them as entirely distinct. The restitution and quest typologies, in particular, overlap and have commonalities. An adapted typology of unresolved quest was identified as the core narrative. Hyden (1997) suggests that the lack of an ending in quest narratives gives rise

to a central problem with respect to illness narratives: they are narratives forever in search of meaning (Hyden, 1997, drawing on Good, 1994). For parents, there was no closure to their quest for diagnosis, although their search and interest in diagnosis commonly reduced over time. Self-transformation was described in that almost all parents came to accept absent diagnosis over time. This transformation was not complete, however, and in many ways the focus on diagnosis did not end but was moved from foreground to background (and iterated between depending on events and feelings) so that a more normal life could be resumed.

Metaphorically, the intensity of their quest for diagnosis had seen them journey from home, spending vast time and energy trying to locate a diagnosis for their child. In reducing their search for diagnosis and changing in their acceptance of absent diagnosis, they return home again. They do not have the precious object they went in search of, instead a different approach to life that, for the most part, accepts the uncertainty of diagnosis and allows them to 'go on'. The puzzle of diagnosis has turned to mystery, an uncertainty they must live with, for now. At the same time, the possibility of diagnosis is kept on, the door to diagnosis left open. Their quest is unresolved, but has been tempered.

This chapter could have ended here. However, the ambivalence of interpretation suggested by the dominant metaphor of stasis means that this chapter can offer no final conclusion or resolution of its own. For if the majority of parents experience self-transformation and were seen to tell a kind of quest story, then why does a metaphor of stasis of movement dominant their accounts? Does the core metaphor of a journey involving stasis tell us that, despite speaking of self-transformation and coming to accept absent diagnosis, parents were more 'sucked into the undertow' (Frank, 1995, p115) of chaos than they indicated? Or simply that the journey was difficult to travel, and that movement on this journey was impeded and often halted? It may simply allow us to know that coming to accept absent diagnosis over time has been a difficult journey.

Does the metaphor of stasis necessarily betray the idea of quest and self-transformation, or instead highlight the extent of the obstacles on the journey? Had I not analysed the metaphors used by parents, my research task would have been easier and the conclusions reached in this chapter more final. Yet, in doing so, perhaps the heart of the parents' stories has become more visible. As Frank suggests (2010), we can never grasp or finalise the whole

story. Perhaps my urge to make the core metaphor and the core narrative identified fit well together is my need to try and control the meaning of the parents' narratives. Relying on typologies too stringently may influence research output in a way that closes off interpretation and doesn't allow the contradictions and ambivalence in participants' narratives to be seen: 'The tendency toward the tidy or conclusive narrative may then become manifest in academic writing' (Thomas-MacLean, 2004, p1649). Here, I resist that urge to control meaning, and will allow the contradiction, if it is a contradiction, to be.

A further research task was carried out beyond considering the kind of stories parents told me and metaphors used in their accounts. In order to further maintain the integrity of the whole narratives, in the process of my data analysis I selected five parent transcripts and created 'poetic transcriptions' (Glesne, 1997) of the entire narratives. Given word limitations, I do not include these poetic transcripts here. I have, however, included one of these 'found poems' or 'data poems' as sometimes described (Butler-Kisber, 2002; Rapport, 2008; Parsons Emmett et al., 2011) as an appendix (Appendix 8): *The Story Lisa Told me While Kelly Ironed her Hair*.

In carrying out narrative analysis by examining the kinds of stories parents told me, the metaphors used in their narratives, and creating poetic transcriptions, I have endeavoured to maintain the integrity of the whole narratives of participants as much as possible within the scope of the thesis and to honour the stories of living without a diagnosis. By including a chapter taking a narrative approach, and by illuminating themes in other chapters by using extensive parent quotes, I hope to evoke a strong sense of individual participants and to reduce the risk of thematic analysis being reductive with participant's words detached from their original context. It is hoped this has makes for more powerful, evocative presentation of analysis. The themes of fracture and deferral, alluded to in this chapter, form central themes in the following chapter, chapter 6, looking at how parents made sense of living without a diagnosis.

**6.1 Introduction**

**6.2 Making sense of fractured expectations and assumptions**

*Non-diagnosis has low cultural resonance*

*The disclosure of non-diagnosis*

*Fractured assumptions about doctors and medical science*

**6.3 Making sense of living with diagnosis as deferred**

*Diagnosis as deferred and being kept on at the clinic*

*Liminality, hope and acceptance*

**6.4 Conclusion**



## 6.1 Introduction

This chapter is the first of four chapters considering the consequences of living without a diagnosis for parents. Later chapters (7, 8 and 9) look at the impact of absent diagnosis on how parents thought about cause and prognosis, access to support and services, and in managing social interaction. In this chapter, I suggest that one of the consequences of absent diagnosis for parents may be difficulties making sense of their experience. Parents may experience cognitive disturbance in the face of a diagnosis not being found for their child. This is a difficult theme to explore, as it is more conceptual and abstract than the other key areas identified as being affected by not having a diagnosis. However, it is felt to be important to explore this theme; it was common to parent narratives, and it is an aspect of absent diagnosis that has not been considered in an in-depth way in other studies with families. Parents were not asked directly whether they had difficulty making sense of not having a diagnosis, although they were asked about their knowledge of undiagnosed disability prior to their experience, how they found out a diagnosis may not be possible, and whether they thought not having a diagnosis had impacted on how they felt about doctors.

As suggested in chapter 1, in considering the consequences of (non-)diagnosis as a separate analytical realm from the process or categories of diagnosis, I risk reifying the distinctiveness of these realms and overlooking their circuitous relationship and the way they depend on each other. In reality, parents do not necessarily experience a definitive end to the testing process, nor a distinctive beginning to an era of 'living without a diagnosis'. Indeed, the testing process lacks closure in the absence of diagnosis. The available tests may have been exhausted, yet there is the possibility of new tests and new knowledge or changes in the child's condition that may lead to diagnosis. The parsing of the parents' experience into two distinct realms, as I have done by structuring analysis as process and consequences, is a way of categorising an experience that may not have such distinct boundaries in the everyday life worlds of the parents. I use the model suggested by key sociology of diagnosis authors (Jutel, 2009; Jutel and Nettleton, 2011; Jutel, 2011a) heuristically. In creating a nosological boundary between the process of diagnosis and the consequences of living without a diagnosis, my aim is to allow a meaningful interpretation of parents' experience, and I believe these two realms are distinct enough to allow this division.

Many parents suggested they had difficulties making sense of absent diagnosis. Absent diagnosis may have fractured their expectations, representing a loss of their assumptive worlds (Kauffman, 2002). Not being able to achieve a diagnosis may represent an epistemic shock for parents, as they struggle to make sense of an experience that challenges their assumptions about the world. In chapter 2 it was suggested that there is a contemporary cultural drive towards ever more specific diagnosis, with a shrinking of the normal (Latimer, 2013) and increasing medicalisation of realms of life previously held to be normal. Within this cultural context, being unable to locate a specific diagnosis for significant disability may cause bewilderment and confusion.

The idea of unattainable diagnosis appeared to cause conceptual difficulties for many parents, and was a feature of 19 parent narratives. The absence of diagnosis was expressed as bewildering, with parents finding it difficult to understand what it was they were dealing with in the absence of diagnosis, resulting in a kind of existential or epistemological anxiety. Sarah's mother, for example, said 'I cannot deal with something that is not there' and spoke of 'grief because of the unattainable, the loss of something which has no substance' (IF, AUS). Cameron's father expressed confusion at being faced with the absence of something that was so tangible in everyday life, he said 'it can't not exist because it has to be something'. Many parents referred to wanting 'to know' and suggested that not knowing was difficult. Calum's father, for example, said that the most difficult thing about not having a diagnosis was 'the not knowing'. Claire's mother made numerous references throughout her interviews to the value of 'knowing'. She said 'it's just not knowing...I suppose like for peace of mind you'd just like to know what it was you child has, yeah... I would like to know, just personally'. Her comment suggests that her desire to *know* relates to an inner need; peace of mind would come about in knowing. Similarly, Eva's mother spoke of seeking to find composure of mind again suggesting the experience of non-diagnosis was psychologically unsettling; 'it may help me to settle my mind'. Non-diagnosis was experienced as an absence, in contrast to the very tangible reality of the difficulties experienced by their child. Diagnosis had an absent presence; significant impairment impacted on their child's life, health and development yet a specific label remained elusive. An incongruence, or sense of absurdity, may be felt; their child's impairment is signified in experience but has no signifier in language.

This chapter will examine different aspects of the way parents made sense of absent diagnosis and look at the cognitive difficulties parents experienced. I explore how parents made sense of absent diagnosis in the context of the fracturing of their expectations and assumptions (6.2), and how they made sense of diagnosis as deferred (6.3).

## **6.2 Making sense of fractured expectations and assumptions**

Here, I consider how parents made sense of living without a diagnosis with reference to a theme of fractured expectations and assumptions. I explore the idea that *non-diagnosis has little cultural resonance* and that this may increase the sense of confusion for parents when faced with a situation they are unfamiliar with. Lack of familiarity with undiagnosed childhood disabilities and no contact with other families living without a diagnosis may contribute to the cognitive difficulties parents may have making sense of absent diagnosis. Their expectations, that significant disability is named by diagnosis, are fractured. I then consider *the disclosure of non-diagnosis*; how parents learn that a diagnosis may not be achieved for their child. If parents experience a moment of disclosure, in which they learn a diagnosis may not be possible, then this may cleave their previously held assumptions. Following on from this, I suggest that the experience of non-diagnosis represents a *loss of parents' assumptions about doctors and medical science* and that these assumptions and expectations may be brought into view by the process of being fractured.

*Non-diagnosis has low cultural resonance*

Very few parents (four) had known about childhood disabilities that were very difficult to diagnose prior to their experience. Parents often knew about commonly diagnosed childhood disorders, but few knew about disorders that could not be named.

I didn't know anything about unknown syndromes or that, mostly just Down's (Claire's mother, 1)

Those parents who had encountered undiagnosed conditions either worked as health professionals (as a doctor, a nurse and a therapist) or had a disabled member of their extended family who did not have a diagnosis. Parents were also asked whether they currently knew other families with disabled children who did not have a diagnosis. Only nine parents reported having contact with other families with children who did not have a diagnosis. This contact in almost all cases had been established since the birth of their own child. In two of these nine cases this contact was established as a result of parents setting up their own support organisation for families living without a diagnosis.

The situation of non-diagnosis may have very little cultural resonance. People may be unfamiliar with rare or difficult to diagnose conditions and may never have encountered the idea of a child having significant and life-limiting impairment with no unifying diagnosis. A significant number of parents made reference to conditions with high cultural resonance when talking about their child not having a diagnosis, referring to other people 'understanding' or 'knowing' these common conditions.

If you say to somebody oh my child's got Downs they can understand and know what that means (...) it is very difficult because if Claire was Down's you'd say Oh she's Downs and a lot of people, the majority, know what that's like they can accept that (Claire's mother, 1)

People are familiar with Down's syndrome they know it (Moy's mother, TI, USA)

In many cases parents framed these conditions as representing the antithesis of the situation of non-diagnosis. Moy's father, for example, compared public understandings of Down Syndrome with the confusion generated when telling members of the public there was no diagnosis for his child's condition:

When you tell someone your child has Down's Syndrome they get a knowing look on their face. When you tell someone we don't know what her problem is we get a lot of dumb looks (Moy's father, IF, USA)

Half of parent participants referred to Down syndrome and/or Autism Spectrum Disorders as representing a contrasting situation to having no diagnosis.

If you can say my child has down's syndrome or my child has Autism the people are immediately clued into a kind of situation there they're not at all with Emily (..) if you say they have developmental delay what does that mean d'you know (Emily's mother, 1)

If you say to somebody my child has down's syndrome there is a general perception out in the community ... about what down's syndrome means and what down's syndrome looks like and what the prognosis is for down's syndrome ... whereas with Jessica she doesn't have a diagnosis (...) there isn't anybody who knows what it's like to be us (Jessica's mother, TI, AUS)

Here, Jessica's mother suggests that Down syndrome is understood in the community, there is a general perception of what it means. Without a diagnosis, however, there is no solidarity with others, nobody who knows what's it's like to *be* them. The public have a perception of common conditions, in contrast with undiagnosed conditions that don't have this cultural resonance.

I suggest that the low cultural resonance of non-diagnosis may contribute to the difficulties parents may have making sense of living without a diagnosis. If they lack familiarity with the idea of absent diagnosis and lack contact with other families living without a diagnosis, the experience of being unable to obtain a diagnosis for their child may challenge their assumptions and cause confusion and difficulties with making sense of their situation.

*Disclosure that a diagnosis may not be possible*

If, as suggested above, parents were commonly unfamiliar with the experience of significant childhood disability for which there is no diagnosis then, at some point in their experience, they must have come to learn that absent diagnosis was a possibility. For most of the parents for whom it was relevant (19) a moment of disclosure was described. The disclosure that a diagnosis may not be found for their child may represent a moment of surprise or shock for parents. An assumption they held (that childhood disability can be diagnosed following a testing period) is fractured in a moment of disclosure. Parents were directly asked about how they came to realise that it might not be possible to obtain a diagnosis for their child. One parent did not respond to this question in the interview. Six parents did not recall a moment of disclosure. Instead they described how it had 'became apparent' over time that their child might not get a diagnosis.

I don't think it was ever really explained as such. It just became apparent (Jessica's mother, IF, AUS)

For those parents who recalled realising over time that their child may not get a diagnosis, this realisation came after months or years of tests and assessments. As time went on, as more and more tests came back negative, or as they reached a situation where there were no more tests to conduct, they came to their own conclusion that a diagnosis may not be available to them.

This wasn't really explained to me but I reached the realisation [that there would not be a diagnosis] when the doctors couldn't suggest any further tests (Peter's mother, IF, AUS)

Most parents (19), on the other hand, recalled a moment of disclosure in which they were told that it might not be possible to find a diagnosis for their child.

She said sometimes you have to be aware that there's not a diagnosis and there's no definite cause and that you know unfortunately is how it is (Emily's father, 1)

Some parents felt the timing of this disclosure made the experience more difficult. The following extracts illustrate that there could be variation in parents' ideas about when this

disclosure should take place, with some suggesting it came too early, and others suggesting it came too late.

I certainly didn't need to hear any of that right at that point (Moy's mother, TI, USA)

I think it would have been good if it had been introduced a bit earlier (Ismail's mother, 1)

I can mind Dr [name] saying there's a lot of children that have difficulties that are unknown and that they're never known...so you may never know what Olivia's condition is...it was like pfff you know I can't put that into words it was just overwhelming (...) I thought how could she say that you know start telling me this when investigations had not even started yet (Olivia's mother, 1)

Ten parents reported that being told there might not be a diagnosis was a negative experience and that this moment of disclosure could have been improved upon, their suggestions most commonly related to the timing of the disclosure. Four parents reported that they were told in a positive way that there might not be a diagnosis, and five that this experience was neither positive nor negative. Several suggestions were made for improving the way that it was disclosed that a diagnosis may not be possible. These suggestions included: more thought about the timing of this disclosure; offering information leaflets or additional information about living without a diagnosis or about why some conditions are difficult to diagnose; and a mentor scheme for parent-to-parent support or contact with other families with undiagnosed children.

I suggest that in making sense of living without a diagnosis, having a moment of disclosure may represent an epistemic shock and be difficult for parents as their assumptions and expectations about receiving a specific diagnosis after a period of testing are suddenly challenged. Yet if undiagnosed childhood disability has very little cultural resonance, it is inevitable that parents will learn at some point in the process of testing that it may not be possible they may not get a diagnosis for their child. For the parents in my own study, the epistemic shock of learning that there may not be a diagnosis may have affected how they coped and came to terms with living without a diagnosis and may have made it difficult to make sense of absent diagnosis.

The experience of not being able to locate a diagnosis for their child may represent a loss of parents' assumptive worlds and may be incongruent with parents' expectations of doctors and medical science. Previously held beliefs and assumptions about the world, and in particular doctors and medical science, may be fractured. Parents were not asked directly about their expectations of health professionals or medical science, but they were asked whether their experience had affected how they felt about health professionals. Many parents said that their experience with their child had affected how they saw doctors and more than half of parents spoke spontaneously about how not having a diagnosis had impacted on their expectations of doctors and medical science, which had previously been high.

Of the 16 parents who reported that their experience had impacted on their expectations of medical science, 10 reported that their expectations of doctors and medical science had been unrealistically high prior to their experience of seeking a diagnosis for their child. Calum's father, for example, said: 'I had a false impression of doctors, that they were the all knowing all seeing oracles. I expected this to happen, a magic label' (2). He described how his expectations were far greater than the actual abilities of medical science. Initially, his high expectations meant he had difficulty making sense of how doctors and medical science could achieve so much in other areas, yet not come up with a diagnosis for his son.

It's like the cloning of Dolly the sheep. They can clone animals. The genetics technology is there to create a human being and organs and yet why can't they tell us? (Calum's father, 2)

He suggested these high expectations were influenced by media representations of medical science and that his expectations had adjusted as a consequence of his experience.

Without a doubt our expectations were far far greater than the actual abilities of medical science just now and that's our fault not the doctor's fault...you see so many wonderful things on the telly with medicine nowadays and you think all of medicine's up there and genetics is it's an amazing subject but it's maybe not quite where we expected it to be...we've got more realistic expectations now (Calum's father, 1)



Calum's mother spoke about how she expected, prior to finding out it was difficult to diagnose her son, that doctors would be able to locate a diagnosis from simple tests: 'I thought they'd do some blood tests or whatever they'll be able to say right this is what he's got' (1). For this reason she felt not obtaining a diagnosis for her son had impacted on the way she saw doctors and medical science.

Abdul's father described how his opinion of consultants had changed as a result of his experience with his son. While initially he may have regarded the consultants involved as having unique knowledge, over time he had come to realise that this knowledge was simply acquired over time and not uniquely available to them.

Initially I think you know you think to yourself yeah oh well these guys know it all kind of thing...but eh subsequently you just think...he's just a guy who's old because he's been doing it for a while...you know that's it the only reason that they're consultants is because they've stayed there a long time and yes they have knowledge clearly but it's not as if you know they're brains on sticks (Abdul's father, 1)

He also described a transformation in his expectations of life more generally as a result of his experience of seeking diagnosis for his son. He spoke of a previous solution-demanding attitude to life, of 'expecting answers', and how the experience of being unable to obtain a diagnosis had challenged this expectation.

Abdul's mother also felt that her expectations and assumptions had changed. She described how the experience of not being able to find a diagnosis for Abdul had an impact on how she saw doctors and medical science, referring to her expectations of doctors prior to the birth of her son.

I think we expect doctors to know everything and we expect doctors to be able to fix everything and we expect them to never have bad days and we expect them you know to em as I say just to fix it...I now realise that you know I think we mentioned earlier it's not an exact science they don't have the answers and that's just life (Abdul's mother, 2)

She suggested a change in assumptions about expecting answers in life more generally.

It was just the sort of realisation that there isn't an answer to everything (...) you realise how much we do expect answers you know and so to basically for someone to say we don't have an answer is quite hard...it's hard to accept...having gone through this experience my expectations have changed (Abdul's mother, 2)

Similarly, Peter's mother highlights both our cultural expectation of doctors and the way her experience had transformed this expectation; 'It has helped me to understand that doctors often don't have answers, although we like to rely on them for these' (IF, AUS). Olivia's mother spoke of the way she had expected doctors to have 'an answer for everything'. She no longer saw doctors as the 'miracle workers' she had previously held them to be.

I mean you put your trust in these consultants and you I don't know what we think that they are miracle workers but you think that they must have an answer for everything, but obviously not I suppose they're only human aye but you just expect them to know everything (Olivia's mother, 1)

A small number of parents also spoke of feeling let down (Emily's father), losing respect for doctors (Jessica's mother) and trusting doctors less or having less confidence in them (Tom's mother, Cameron's mother and father).

Five parents reported that they did not feel the experience of living without a diagnosis had affected how they saw doctors or medical science. Claire's mother, for example, said she had always realised doctors did not know everything ('because I knew they're no miracle workers', 1) and her experience had not changed that. Of these five parents, three parents who worked in health care said they were already aware of the uncertainties and gaps in knowledge in medicine.

I suggest that high expectations of doctors and medical science may contribute to the difficulty making sense of their experience when a diagnosis cannot be found. Expectations about what doctors know and what medical science can achieve are fractured and, following the loss of previously held assumptions, many parents need to re-evaluate their knowledge and assumptions about the world in order to make sense of their experience.

### 6.3 Making sense of living with diagnosis as deferred

In this section, I look at how parents made sense of living with diagnosis as deferred. It was suggested in chapter 4 that, for parents who consulted with the geneticist, diagnosis was often played as deferred rather than absent. Parents commonly perceived genetics as an advancing field, offering the promise of diagnosis in the future. Here, I consider further this theme of *deferral* in the context of how parents made sense of living without a diagnosis. Here, I extend earlier discussion by including in analysis parents who did not necessarily perceive their child to have a genetic disorder and by looking in a more in-depth way at one parent's account (Eva's mother) of consultation with the geneticist to more fully illustrate the theme of deferral. I also consider the way families were kept on at the clinic, and the meaning of continued clinic attendance for parents.

I start this section by building on earlier work in chapter 4, by looking at the theme of *diagnosis as deferred* in more detail and considering how diagnosis as deferred means that families are kept on at the clinic. I suggest the importance of *being kept on at the clinic* for families. I then consider the lack of closure that may be felt by parents in the absence of diagnosis and the space of *liminality* that may be occupied by parents experiencing deferred or absent diagnosis. How this lack of closure impacts on parents *hope and acceptance* is then considered. It may be more difficult for parents to experience acceptance of their child's condition in the absence of diagnosis, but at the same time lack of diagnosis may permit hope. For the fathers in the study, this hope was tied to the hope for future treatment for their child.

#### *Diagnosis as deferred and being kept on at the clinic*

Many parents (18) suggested diagnosis as deferred, 15 of whom had on-going contact with the geneticist. Parental attitudes to diagnosis were often complex, and at times parents oscillated between describing diagnosis as likely, while at other times unlikely; or suggesting diagnosis as deferred, while at other times suggesting diagnosis as not possible or not likely to be possible. As parents considered living without a diagnosis, and told the stories of their

diagnosis journeys, there was ambivalence in their narratives as they made sense of their experiences. However, it was possible in the narratives of a significant number of parents to identify a strong and recurrent theme of diagnosis as deferred.

As suggested in chapter 4, a significant number of parents perceived genetics to be on the brink of significant change and diagnosis as deferred relied on the notion of medical knowledge as ever progressive. Beliefs about the deferral of diagnosis were bound to the notion of progress, genetics 'moving on' and knowledge improving, and improvements in equipment and testing. Time stood in the way of achieving diagnosis, and diagnosis was often seen as deferred until equipment and tests became available or improved.

I think if we could travel forward 50 years and maybe in 50 years time...we'll look back and they'll be able to diagnosis what Calum has got just no problem but I think it's just medical knowledge just now to be honest' (Calum's father, 1)

They're still searching for a more reliable test (Moy's father, TI, USA)

In Eva's mother's accounts of consultation with the geneticist, how diagnosis was played as deferred is well illustrated (all of the quotes in this section are from our telephone interview, ENG). Eva's mother consulted with the geneticist at a clinic. She described how the geneticist had indicated from their first consultation that he believed Eva had a genetic condition. She recalls how he looked for relevant dysmorphic features at this first consultation; 'he sort of took lots of measurements of finger length and eye distance and all those sorts of things', with the aim of locating a diagnosis for Eva; 'he said that he puts them all together and normally they'll point him towards a syndrome that he knows something about'. However, a diagnosis is not reached and the family return for their second consultation.

I did go back and see him again and he said to me he looked at her again and he said I'm still sure that she has got some sort of chromosomal abnormality he said but I'm almost certain that I'm not going to find anything from the tests that we can look at the moment

The geneticist predicts that he is unlikely to 'find anything' from tests currently available, suggesting the possibility of diagnosis in the future but not now. The geneticist has been unable to recognise a pattern to Eva's features and to fit her into a syndrome he can test for;

'he said the trouble with Eva is that she doesn't fit in to anything that he knows that he can test for'. Eva's mother suggests that inadequacies in the current levels of technology may be responsible for lack of diagnosis.

The clinical geneticist...told us that although the precise cause could not be determined we were informed that it was highly likely that the cause of Eva's disability was a chromosomal abnormality that, with the current level of technology, it is not possible to detect

Eva's mother takes on the notion of deferral of diagnosis and makes reference to deferred diagnosis throughout her interview. She also makes reference to the way that Eva is kept on at the clinic, despite there being 'no further work' planned in the short term. She reports that the geneticist has indicated that in several years, a diagnosis may be possible if the field of genetics has 'moved on'.

No further work is being done or is planned re a diagnosis for Eva. The closest we have come is a suggestion from the Clinical Geneticist that we revisit him when Eva is about 12, really to see if technology has moved on enough to provide any new tests for Eva

Diagnosis is deferred; the possibility of diagnosis remains open, likely even. Eva was 7 at the time of the interview study. At the time of writing this thesis genetic technology has moved on, as anticipated by the geneticist consulting with Eva and her family, and new testing techniques like whole exome sequencing are potentially available. At the time of the interview study, 'the current level of technology' was not able to offer a diagnosis for Eva. Following her consultations with the geneticist, Eva's mother continues her work in trying to recognise a pattern between her own daughter's clinical symptoms and that of other children she finds online as she continues to seek a diagnosis. Both herself and the clinical team continue looking for Eva.

The parents who did not believe that their child had a genetic condition, yet suggested diagnosis as likely in the future (3 parents), believed a diagnosis would be possible if there were improvements in the way the search was being carried out or there was increased funding to make further assessments possible. For the parents who perceived their child to have a genetic condition or who maintained ongoing contact with the geneticist, most (15)

suggested diagnosis as deferred, with the future possibility of diagnosis depending on genetic knowledge advancing.

In the circumstance of diagnosis being deferred, families described being 'kept on' at the clinic (Latimer 2007a, 2013). Around a half of parents suggested it was important that they had continued access to the clinic. Parents appreciated an open referral or continued attendance even if this was reduced in frequency, and knowing they could make contact if their child experienced a change or if new testing was available. With their child kept on at the clinic, the door to diagnosis was still open.

All parents for whom it was relevant (23) continued to attend consultations with the geneticist or other specialist involved in the care of their child. Parents reported continued attendance at specialist clinics with their child, either regularly with planned consultations on an as needed basis, or both. Around half of parents spoke of the benefits of continuing to attend the clinic with their child. When parents spoke spontaneously of their experience of attending the genetics clinic or clinics with other consultants involved in the care of their child, they were asked what they felt they gained from attending the clinic. Parents responded suggesting a variety of reasons and benefits for continuing to attend the clinic with their child, not limited to just medical benefits.

Continued attendance at the clinic offered reassurance for some parents, that everything was still being done for their child and that if any new tests came up or they had any suggestions or questions there was somewhere they could raise these. For some parents, the medical benefit was secondary to the emotional support and connection provided by continuing to attend consultations.

There isn't really anything medical we get out of it you just want some sort of connection there I think, you need to feel there's somebody there, I guess as well because every wee while there are questions that come up for us and it means that you know that there's a way you can someone you can go and ask (Abdul's mother, 2)

Nine parents described encounters in which it was suggested or proposed that their child be discharged from the clinic. On all occasions the discharge was suggested by the clinician/s, rather than the parents. Olivia's father, for example, describes an encounter in the genetics

clinic where it is suggested that his daughter is no longer seen at the main children's hospital by the genetics team, but is instead seen by the paediatrician at the family's local hospital:

They started mentioning so are you quite happy coming here because it's quite a journey for you and em we realise that you know that this'll put you quite out quite a lot having two children and you know [speaking to Olivia's mother] your husband's obviously here taking time off his work and would you like us to just put you back in the community and it was just like you know...suddenly they were just willing to sort of wash their hands...and there was just no way I was settling for that...I wasnae accepting that (Olivia's father, 1)

Olivia's father is not happy to be referred to someone 'back in the community' and wants his daughter to be kept on at the genetics clinic at the children's hospital. A similar suggestion of discharge is recounted by Cameron's father. When the paediatrician his son sees suggests that Cameron is discharged, his father refuses: 'I wouldnae let him' (1). As a result of his refusal to have Cameron discharged, he reports that Cameron is seen by the paediatrician every six months.

Eva's mother also described a consultation during which discharge from the paediatrician's clinic is suggested. As suggested earlier, the clinical geneticist plans to see Eva again in several years and has gone as far as he can with the process of diagnosis for the time being. Here, Eva's mother recounts the suggestion that Eva is discharged by the consultant paediatrician, who sees Eva bi-annually.

At the end of the appointment she [consultant] said to me well maybe she said we could sign you off now and I think she looked at the expression on my face and said oh no perhaps it would be better for me to keep you on my books so we'll see Eva again in a year's time and I thought that really sort of sealed it for me that as far as she's concerned there is no nothing more. (Eva's mother, TI, ENG)

For Eva's mother, the suggestion that Eva is discharged signifies that there is 'nothing more' that can be done, as well as has implications for future contact and potentially for therapies for Eva. I ask Eva's mother how she feels about this.

I was really quite panicky at that point I thought you can't do this it would just be like casting her aside you know she's a very nice she's a lovely woman and we're very lucky in that you could ring her I could ring her up anytime if something serious did happen and just request another appointment you know I have done this a couple of times in the

past...and the thought that she might just suddenly sign me off and I thought if that happens it might mean that I'm not going to get occupational therapy or physiotherapy or anything and it's just like you sort of feel like you're being set adrift really (Eva's mother, TI, ENG)

The clinic provides an anchor for Eva's mother, without which she feels she may be 'set adrift' with her daughter 'cast aside'. This indicates the possible intertwining of the medical into the everyday worlds of families, and the importance of that presence. The medical contact anchors the experience and this continued contact with the consultant may assist coping with or making sense of absent diagnosis.

Emily's parents also experience suggested discharge for Emily, but for them discharge to the local clinic goes ahead and they describe positive consequences of the transfer to the local clinic. They recognised that there was 'no point' in continuing to go to the genetics clinic so frequently when all the tests were normal. They are transferred to a paediatrician at their local hospital on the understanding they will meet with the paediatrician every six months, and both the paediatrician and a geneticist every year or two in a joint clinic. The family seem happy with this arrangement, with Emily's mother feeling it maintains the level of contact they need; 'medical science is always advancing...so that leaves the door open...it allows you a medical contact.' For Emily's mother, it is important that they are not completely discharged, which for her would mean being left without an important contact:

[if discharged from the clinics] we would be swimming about with nowhere to go...with no-one to throw you a lifeline...if you had a difficulty then at least you would be able to talk to someone about that (Emily's mother, 2)

Although Emily's parents experience being discharged from the genetics clinic as positive, perhaps because they have on-going contact with the geneticist despite being kept on at the local rather than regional clinic, the majority of parents who reported suggestions their child was discharged from the clinic experienced this as negative. Appropriate on-going medical contact may be of great value to families living without a diagnosis. The importance of being kept on at the clinic was suggested by around half of parents. With their child kept on at the clinic, the door to diagnosis was not closed. Furthermore, the benefits of continued attendance at the clinic were not just medical, but on-going support was also provided in terms of living without a diagnosis. In this way, the realms of the process and consequences



of diagnosis exist in tandem. On-going clinic attendance means on-going care, both medical care and pastoral care as parents are supported through their continued journey.

### *Liminality, acceptance and hope*

In exploring the process of diagnosis for parents in chapter 4, it was suggested that perpetuity was a feature of this process and that, while parents may have reduced their search activity and focus on diagnosis, this process does not end. With an absence of closure, parents may occupy a space akin to liminality (Turner, 1987). In chapter 2, I introduced ethnographic studies of dysmorphology, in which it was suggested that parents in the paediatric genetics clinic occupy 'a space of motility' (Latimer, 2007a, p126) in their participation in the clinic, shifting back and forth between deferral and definition as they negotiate potential diagnosis. This motility in which they are held is not felt to be the same as liminality, for parents in the dysmorphology clinic are moved between these states through participation not sequestration or subjection (Latimer, 2007a, 2013). Several parents in my study referred to being 'in limbo' and a number of parents spoke of being unable to 'move on' and referred to a lack of closure. David's mother, for example, said 'it's very stressful because it's not a final it's not finalised and I think there should be a finalisation of something...it's continuous' and made repeated references to wanting to 'move on'. Jessica's mother said that living without a diagnosis was like 'embarking on a journey that has taken some unbelievable twists and turns and shows no sign of ending', while Moy's father said simply, 'it doesn't end'.

In their space of diagnostic limbo, I suggest that parents occupied a space akin to liminality, a no man's land between problem recognition and diagnosis. As they reside in this space in the long term, potentially perpetually, this is not the same as liminality where one passes through and is only temporarily at threshold, betwixt and between (Turner, 1987). The ritual of diagnosis may never be complete. Liminality, according to Turner (1987), must eventually dissolve. It is an intense state that cannot exist in the long term without some sort of structure to stabilise it, either by return or normalisation. Parents may resolve the felt need for closure, and an end to their state of limbo, in a number of ways; they may move on, reducing their search for diagnosis and building uncertainty into their lives in this way; they may adjust their previously held expectations and assumptions about doctors, medical

science and the world; they may continue to attend the clinic and maintain hope for a diagnosis in the future, while allowing their thinking about diagnosis to occupy a background rather than foreground place in their lives.

With reference to acceptance, parents were asked directly whether or not having a diagnosis had impacted on their ability to cope with or adjust to their child's disability. Many parents were not able to answer this question or said they were unsure whether or not having a diagnosis had made a difference to their acceptance or coping with their child's disability. This is not unexpected given that parents did not have an understanding of how they would have coped if they *did* have a diagnosis as they had not experienced this, as Eva's mother highlights in her response.

It's been a long drawn out process. I can't really say, because I haven't experienced having a diagnosis. However the other day I heard a mother of a girl with Down syndrome saying that it really took her 6 months to come to terms with her daughter's diagnosis. Maybe she is a much stronger person than me, because Eva is 7, and I still haven't come to terms with things (Eva's mother, IF, ENG)

Ten parents said that not having a diagnosis had impacted on the way they had coped with or adjusted to their child's disability. Several parents referred to the prolonged nature of the period of testing or the long process of seeking a diagnosis, suggesting this aspect of living without a diagnosis impacted on coming to terms with their child's disability.

It's a really painful long drawn out process if if some if you could have a diagnosis if somebody could just say to you blah this is what you've got this is what your child has we've done a test and it's conclusive this is what you have you can well you grieve of course and it's devastating and all the rest of it but then you can move on whereas if you don't have a diagnosis you're almost almost like in limbo in a lot of ways you can't move on [sighs] (Jessica's mother, TI, AUS)

We both found it hard to accept Peter's disability when we didn't have a name for it...your life seems to revolve around trying to find out what it is so instead of just accepting and getting on with it you feel like you are constantly searching for information and answers...it has taken us longer to adjust to the disability...sometimes we are full of hope and sometimes we feel completely desperate (Peter's mother, TI, AUS)

Here Jessica's mother refers to a grieving process associated with discovering your child's diagnosis, suggesting this is not available in the absence of diagnosis and so you remain in a

liminal state unable to move forward, while Peter's mother describes oscillating between hope and despair in a life revolving around trying to find a diagnosis rather than accepting the disability and getting on with it. Calum's father, like Jessica's mother, makes reference to a process of adjustment that may be absent when your child doesn't have a diagnosis. He believes that not having a diagnosis affects the 'initial coping mechanism' that would allow parents to adjust to their child having a disability. This coping mechanism is perceived to be present for parents of children with named conditions, but absent for parents of children who do not have a diagnosis.

Perhaps getting a diagnosis would have helped move on but we didn't get that unfortunately so (...) I think realising that your child's got special needs will probably equate to coping with bereavement...it's a bad comparison but when somebody dies...you know you've got to cope with it and there's mechanisms there to help people get over grief or whatever bad things happen in their life I'm no saying having a special needs kid's bad by any stretch of the imagination but it's different if you had a diagnosis I think that that's there's a mechanism there already it does yeah [affects your ability to move on] because you know you're dwelling on it (...) the main one [impact of not having a diagnosis] is without a doubt being able to the initial coping mechanism yeah (Calum's father, 1)

Two parents did not feel that not having a diagnosis had impacted in this way. Anna's mother, for example, said in response to being asked if not having a diagnosis had an impact on how she coped with or came to terms with her daughter's disability; 'No she will be what she will be' (TI, USA) and felt her Christian faith had helped her to cope with her daughter's disability.

Absent diagnosis may be seen to impede acceptance for parents, yet at the same time allow for hope. With reference to hope, many parents (17) expressed hope that a diagnosis may be possible in the future. Six parents expressed hope in the context of speculating that a future diagnosis may offer the possibility of treatment or cure for their child. The six parents who expressed hope about treatment were all fathers; none of the mothers expressed hope for treatment or cure in this way. Emily's father, for example, was convinced that 'as medicine has been progressing over the years' a diagnosis would be found for his daughter one day; 'a few years down the line they might know quite clearly what the problem is...we never give up hope.' Although he felt he had accepted his daughter's difficulties ('we accept it as the way it is') he referred to the possibility for treating her difficulties if a diagnosis should be

found; 'I'd like them to know what the problem is. It'd be even better if they could do something about it, but we accept Emily just the way she is.'

Similarly, Cameron's father felt that his son would get a diagnosis someday, and that diagnosis may open the door to help for his son.

If he got re-tested or re-assessed for these things they may get different results and there could be something there that could help Cameron, whether it be slight or whether it be major you know, who cares as long as it helps him (Cameron's father, 1)

Although he felt there may be something that could help his son in the wake of a clear diagnosis, he doubted a cure was likely. Yet he still held onto hope that some improvement may occur if a diagnosis could be found.

I've resigned myself the fact that there's no going to be a total cure, I mean albeit I would like Cameron to be perfect for want of a better word....it's just in case there's some new test that would help Cameron...whether it be you know some kind of treatment or therapy you know that could help Cameron write better or speak better or do anything better be more independent in any way (Cameron's father, 1)

Olivia's father felt that his 'head' was telling him it was increasingly unlikely that his daughter would get a diagnosis, although his 'heart' was telling him that they would 'find something'. Like the other fathers, he linked obtaining a diagnosis with the possibility for treatment, even while realising that his hopes may be unrealistic. He felt that his daughter could not be 'the only one on the planet that has this' and that there must exist 'others out there who are being treated for this' (1).

That's the great hope I mean I think certainly for me, in an ideal world or whatever, if you like...the sort of happy ever after ending is that we get diagnosis, we go down the path that diagnosis points us to go down, and then there's some sort of medical development, whether it's gene therapy or something that you know we can start repairing or nurturing what needs to be nurtured to bring Olivia on...that's the heart hope isn't it, the head hope you know realistically we might never get there but I think you've got to get up in the morning and have something to cling on to (Olivia's father, 1)

Calum's father, also keen to point out that his hopes may not be realistic, hinted that at some level he held out for treatment for his son. He suggested there were times he had visited his

son's consultant and wondered whether they could 'give him this drug that might maybe not cure him but help him...bring him on that little bit more' (1). Moy's father also suggested that his hopes about diagnosis were also hopes about treatment for his daughter; 'a diagnosis could lead to more aggressive treatment based on other similar patients' (TI, USA). Abdul's father described hopes for cure in the past, as he speculated about stem cell therapy. Although realising that a 'magic bullet' cure was not possible, he did not entirely rule out the idea of cure.

There are no real magic bullets as such I don't think so, and I'm not sure that em we can find one, as I say I had a fanciful about stem cells cause I'd read about them thinking oh well if he's got a gap in his brain maybe maybe we can fill it in with stem cells and then magically make it all better (Abdul's father, 1)

Six parents (all fathers) expressed hope that diagnosis may lead to treatment that offered possibilities for improvement. In the absence of boundaries about what to expect, this hope could be kept alive. Hope for treatment and cure was not identified in any of the mothers' narratives, although many hoped for a diagnosis in the future.

In this section I have suggested that without diagnosis parents occupy a space akin to liminality. Unlike Turner's (1987) liminality, however, parents are active in negotiating potential diagnosis and thus occupy the liminal state through participation not subjection. This draws on earlier work on parents in the dysmorphology clinic (Latimer, 2007a, p113; Latimer 2013). I further suggest that parents act to move beyond this liminal state by reducing their search for diagnosis, accepting uncertainty (to varying degrees) and consigning absent diagnosis to the background rather than the foreground of their thinking and lives. Yet, at the same time, the possibility of diagnosis is kept open, and many parents expressed hope for a diagnosis in the future. Fathers expressed hope differently to mothers, in that all but one father hoped for treatment that would offer improvements for their child. With regards to acceptance, ten parents felt not having a diagnosis had impacted on their acceptance of their child's condition, although parents found it difficult to respond to this question.

## 6.4 Conclusion

I have suggested that how parents made sense of living without a diagnosis can be understood in the context of the themes of fractured expectations and assumptions, and diagnosis as deferred. As non-diagnosis has low cultural resonance, and most parents were unfamiliar with undiagnosed childhood disability prior to their experience, they may have experienced a fracture of their expectations and epistemic shock that diagnosis was not possible. This may have been compounded by finding out in a moment of disclosure that a diagnosis may not be found. Parents' assumptions about the world, in particular their expectations of health professionals and medical science, were potentially fractured by the experience of absent diagnosis in the face of significant disability. Kelly (2009) suggests that the birth of a disabled child for some parents can be experienced as an epistemic shock, exposing the limitations of medical knowledge and practice (Kelly, 2009, p94). I suggest that for parents of children with significant disability yet no diagnosis, this epistemic shock may be more acute. The parents' assumptions, for example that medical science would be able to provide them with answers, come into view as they are challenged. I suggest non-diagnosis is a natural breaching experiment (Garfinkel, 1967) exposing our assumptions about the world. Parents may have difficulties making sense of their experience in response to the epistemic shock of absent diagnosis.

Parents described conceptual difficulties associated with living without a diagnosis. Other research with families has suggested difficulties associated with uncertainty of diagnosis including the possible impact on coping and adaptation and the cognitive disturbances that may occur in the absence of a certain diagnosis, with parents strongly feeling a need 'to know' and feeling frustrated and unsettled by absent diagnosis (Rosenthal, Biesecker and Biesecker, 2001; Lewis, Skirton and Jones, 2010; Graungaard and Skov, 2007; Madeo et al., 2012; Lenhard et al., 2005; Makela et al., 2009; Genetics Alliance UK, 2010).

The uncertainty that non-diagnosis brings can result in confusion (Brookes-Howell, 2006). Uncertainty is identified as an important concept in understanding the impact of a condition on parental adaptation (Stewart and Mishel, 2000) and uncertainty is understood as a major component of the illness experience that affects psychological adaptation (Lipinski et al., 2006). Skirton (2001, cited in Brookes-Howell, 2006) suggests that clients in the genetic

counselling clinic require certainty to prepare for the future and to understand the present, and for peace of mind. Applebaum and Firestein (1983) recognise the impact that non-diagnosis can have on the parental role, with non-diagnosis resulting in a unique vulnerability that, along with feelings of helplessness and fear of the unknown, can disrupt parental adjustment to the child's problem.

I suggest that the experience of parenting a child without a diagnosis may represent a loss to parents' assumptive worlds. Janoff-Bulman's (1992) assumptive world theory is a cognitive theoretical perspective for understanding the negative impact of traumatic events based on a principle of shattered assumptions about ourselves and the world. The theory suggests that individuals under normal circumstances hold positive assumptions about themselves and the world based on belief in a just, meaningful, and benign world. One of the negative effects of traumatic life events is the shattering of these fundamental assumptions (Janoff-Bulman, 1992), and a major task of recovery is the re-establishment of viable beliefs as a framework for understanding oneself and the world. Parents, on learning that a diagnosis may not be possible for their child, must readjust their expectations of doctors and medical science. In the meantime, absent diagnosis may be experienced as bewildering, as previously held assumptions are fractured.

Furthermore, the loss parents experience may be ambiguous. An ambiguous loss is a loss that remains unclear, characterised by lack of closure or clear understanding (Boss and Couden, 2002). The most stressful losses are those that are ambiguous and humans have the ability to cope with stressful situations as long as they can understand their situation in meaningful ways (Boss and Couden, 2002). Ambiguity in illness states can block this understanding presenting a challenge for coping; the family can't move on and the ambiguity surrounding the condition keeps people confused (Boss and Couden, 2002). It has been suggested that ambiguous loss can be experienced by parents of children with Autism Spectrum Disorders (O'Brien, 2007) and can be characteristic of the experience of parenting a child with significant disabilities (Patrick-Odd and Ladd, 2010).

The low cultural resonance of non-diagnosis is suggested by parents' lack of awareness of undiagnosed disability prior to their own experience. Conditions with high cultural resonance, Down syndrome and Autism Spectrum Disorders, were often framed as

representing a contrast with their own situation of absent diagnosis. The majority of parents recalled a moment of disclosure in which they were made aware that they may not get a diagnosis for their child, representing a moment of fracture in parents' knowledge and assumptions. This loss of their assumptions and expectations, in particular about doctors and medical science, may contribute to cognitive difficulties parents may have as a consequence of living without a diagnosis.

Many parents suggested diagnosis as deferred. Tests were not available *yet* that would lead to diagnosis, but medical knowledge was improving all the time. An important aspect of deferred diagnosis was that families were kept on at the clinic. Literature examining the process of paediatric genetic diagnosis describes dysmorphology as a field of uncertainty in which deferral rather than doubt is performed (Latimer 2007a, 2007b). There is significant uncertainty associated with identifying a new or complex genetic variation (McLaughlin and Clavering, 2012) and the classification of genetic diagnosis is 'as much about doubt, and the precarious nature of genetic diagnosis, as it is about certainty' (Latimer 2013, p8). There are not yet any specific tests for many syndromes (Latimer 2013, p89), so despite the extraordinary disruption that many children experience in their development, a mutation at the molecular level has not yet been identified (Latimer, 2013). As a new frontier of knowledge, genetics is committed to a future in which that which is currently not visible becomes so (Latimer, 2007b). The dysmorphology clinic emerges as a 'space of deferral' (Latimer et al., 2006) and a rhetoric of improving technology is employed as diagnosis is deferred. Parents, like clinicians, may be committed to future scientific advances and the belief that advancement in science that will provide more answers about their child's condition (Reiff et al., 2012). In the absence of certain diagnosis, families are 'kept on' at the clinic (Latimer 2007a p106; Latimer 2013). For parents in my own study, being kept on at the clinic had benefits beyond the medical needs of their child, and a door was left open for support, advice and for future diagnosis. Being kept on at the genetics clinic can have other benefits for families, for example the clinic can be a site of reassurance (Latimer, 2013), a confessional space in which much 'sentimental work' is carried out (Featherstone, Gregory and Atkinson, 2006). In remaining in a space of deferral, parents enact a kind of liminality. The perpetuity of the diagnosis process may represent a kind of no man's land for parents, as they occupy a liminal space of 'watchful waiting' (McLaughlin and Clavering, 2012, p460, drawing on Aase, 1990). Yet they may have motility in this liminal space in the way they



actively shift between seeking definition and accepting deferral or absence and the way they stabilise this liminal state by allowing a focus on diagnosis to move between foreground and background, so that a new normality is possible. In this new normality diagnosis is deferred. Parents may continue the detective work of trying to solve the diagnosis puzzle and, while their search activity and focus on diagnosis may reduce over time, it may not end.

## **Chapter 7      Unknown Aetiology in the Absence of Diagnosis: Self-blame and Risk of Recurrence**

### **7.1      Introduction**

### **7.2      Self-blame**

*Mothers' self-blame*

*Fathers' self-blame*

### **7.3      Risk of Recurrence**

*Impact on Future Reproductive Choices*

### **7.3      Conclusion**

## **7.1 Introduction**

As already suggested in the introduction to the thesis, absent diagnosis had an impact on how parents conceived of the past, the future and the present. In this chapter, I begin by considering the temporal realm of the past in looking at one of the key areas that parents perceived not having a diagnosis had an impact on; knowing what caused their child's difficulties. All parents speculated about what caused their child's difficulties. Seven parents spontaneously raised issues relating to cause when telling their story at the beginning of their interview, or when responding to my question about whether diagnosis had an impact on their lives. The remaining parents talked about issues relating to cause when prompted by my question about whether they thought not having a diagnosis had an effect on how they thought about what caused their child's difficulties.

None of the parents knew with certainty how their child's disability was caused. Without a diagnosis, certainty about aetiology is not possible, although for many a genetic cause was suspected. Most parents speculated about whether they thought their child's condition was genetic, with only 4 parents ruling out genetic diagnosis and a further 3 parents for whom it was not relevant. All of the parents reported that not having a diagnosis had an impact on the way they thought about what caused their child's difficulties. This chapter will look at the way parents experienced unknown aetiology in the absence of diagnosis; in particular how they expressed self-blame when thinking about cause, and the impact of absent diagnosis on knowing the risk of recurrence. In doing so, I consider how absent diagnosis made it difficult for parents to make sense of the past, in that they did not know what had caused their child's disability. Not knowing what caused their child's disability had an impact on how they perceived the future, and for many parents affected future reproductive choices.

## 7.2 Blame and Responsibility

All parent participants reported seeking a reason for their child's difficulties, with some describing an intense, prolonged and persistent desire to know what caused their child's disability. Many of the mothers (16 of 19), and one father (of 7) described considering whether they were to blame for their child's difficulties. Parents speculated about a range of causes for their child's disability. These included causes originating in the prenatal period or before conception (stress, alcohol, nutrition, health and fitness, infection, the mother's age, chromosomal/genetic factors, or other environmental factors); the perinatal period (difficulties in labour, oxygen deprivation, meconium aspiration); and less commonly early childhood (injuries, environmental factors). Parents were not asked directly about self-blame; those who described feelings of self-blame did so spontaneously when talking about cause. Although for some parents speculating about self-blame did not appear to have had a significant impact on their lives, for others speculating about whether or not they were to blame for their child's disability was an on-going or recurring issue that they talked about as having a significant impact on their lives. There were variations in terms of proportions of mothers and fathers who expressed self-blame, with many more mothers proportionately expressing concerns they had done something to cause their child's difficulties.

### *Mothers' self-blame*

All of the mothers speculated about what may have caused their child's disability, with most mothers (16) speculating about whether they were to blame for their child's difficulties. These mothers expressed concerns about risk behaviors before or during pregnancy, during labour and, less commonly, in their child's early years. They worried about what they may have done or not done that may have caused their child's disability.

Did I do something wrong in pregnancy because I'm not a good eater? (Kamil's mother, 1)

My father died suddenly when I was 8 weeks pregnant and I was with Mum when she found him. It was a very stressful time...yes we still wonder if it's something I did in pregnancy. (Peter's mother, TI, AUS)

Mothers often speculated about more than one possible cause, in the absence of a diagnosis suggesting a specific aetiology. Cameron's mother, for example, considers the stress of her house move and the impact of doing too much at this time, along with a decision to defer going to hospital while in labour.

We moved house and I thought you know maybe I've done more than I should have...I sort of think well maybe I left it too late that morning to go to the hospital (Cameron's mother, 1)

Abdul's mother speculates about multiple possible causes of her son's disability, including something being 'wrong' with her, stress at work during pregnancy, drinking small quantities of wine and choosing to eat non-organic food during pregnancy.

You start thinking is it something that's wrong with me? Is it something that happened to me while I was pregnant?...I was worried about it because when I was pregnant it was quite a stressful time at work and so initially I thought is it my fault?...because you don't know there's always that wee bit in the back of your mind...you think well maybe I did have you know a couple of glasses of wine at that stage...I'm going back through all that again in terms of what I did do or what I did not do, was it because I didn't eat organic food you know?...maybe this problem is because of all this stuff that we eat? (Abdul's mother, 1)

Later in her narrative, in her second interview, she speculates about over the counter medication.

I took an anti-histamine...and I didn't realise you weren't meant to take them when you were pregnant and I had taken it ... and I was sort of thinking right what are the effects you know...what could this have caused (...) I was worried about it (Abdul's mother, 2)

Mothers looked back to their pregnancy, labour or their child's early years (for those whose child's difficulties were not apparent at birth) and speculated about a range of factors; genetic, behavioural and environmental, trying to understand what they may have done 'wrong'. Common concerns were toxins in pregnancy, being too stressed, not eating the

right foods, and decisions made during labour. In questioning what may have caused their child's difficulties, they raised questions that remained unanswered.

I mean did I do something wrong? Did I not should I have gone back to work and put him in day care where he'd be with other kids? Should I not have had a drink you know in pregnancy? I mean I didn't drink in pregnancy but I had the odd glass of wine, maybe that's what did it? Maybe he fell and bumped his head you know when he was such and such an age? (Rupert's mother, 1)

Even when the mother strongly suspected, or had been told by their child's consultant, that the condition was likely to be genetic, they could still express self-blame, particularly if they felt the 'faulty gene' came from their side of the family or that they may have done something to 'damage' their genes. Calum's mother worried that her side of the family ('Is it my side? I feel guilty', 1) might have been responsible for the defective gene that potentially caused her son's disability. Similarly Claire's mother was sure that her daughter's disability was caused by something 'in the family', and said 'I've always blamed myself for the way she is' (1). She suspected that her daughter's condition originated from her side of the family as other family members had difficulties; 'it's definitely my side of the family...I'm about 90% positive that it's my side' and expressed guilt that her side of the family was potentially to blame.

Eva's mother worried that her exposure to radiation at work before her pregnancy had caused her daughter's disability.

I have constant worrying about the cause, particularly if it occurred during pregnancy or preconception, was it my fault? I worked in [name of department] before I was pregnant and I am haunted by the suspicion that exposure to radioactivity may have caused her disability... I still feel guilty. I keep thinking, what have I done? (Eva's mother, IF, ENG)

Despite reporting being told by the geneticist that her daughter's disability was likely to have a genetic cause, Eva's mother still expressed a need to know about aetiology and guilt about potentially causing damage to her chromosomes.

I said to him could that have happened preconception or post conception?...I was saying can you say whether it was anything to do with me or not? (...) I still feel

guilty because I kept saying well what have I done that I damaged my chromosomes? (Eva's mother, TI, ENG)

Around half of the mothers (9) specifically talked about 'guilt' or 'feeling 'guilty' suggesting negative feelings associated with speculating about whether they were to blame for their child's disability: 'I still feel a lot of guilt for her low birth weight' (Anna's mother, IF, USA); 'For a while we wondered how it happened I feel guilty' (Calum's mother, 1); 'I couldn't find out what caused it, and I think that's part of the guilt thing really' (Eva's mother, TI, ENG). Five of the mothers, when describing their conduct in pregnancy, labour or their child's early years made reference to the way they 'should' behave.

I did smoke when I was pregnant yes but I never drunk any alcohol I never took any tablets or anything like that, but you just feeling because you're the one who's carrying them you should be the one that's protecting them and when they've got something wrong [silence] (Claire's mother, 1)

Three mothers; Ismail's mother, David's mother and Emily's mother, reported that they did not speculate about whether they were to blame for their child's difficulties. Ismail's mother was not the birth mother of her son, but instead his sister (with parental guardianship) and so she did not express self-blame in relation to the pre-natal or pregnancy period. She did, however, speculate about aetiology and wondered whether her son's difficulties were caused by his lack of oxygen at birth.

When he was born cause he wasn't breathing you know he had his cord wrapped round his neck quite a few times...I do think well maybe it was that...you know I do wonder what caused it (Ismail's mother, 1)

David's mother was convinced that her son's difficulties were caused by her contracting food poisoning when pregnant, even though it wasn't possible to have this confirmed. She described having 'a mother's instinct' during pregnancy that her son was going to have difficulties caused by the infection. She also explained that there was no other possible reason for his difficulties. She described how she had ruled out genetic causes in her mind, as there was nothing previously 'in the family', and also ruled out pre-natal causes. As she had avoided risk behaviours in pregnancy, she felt she could not possibly be culpable for her son's disability.

I think because there was no other explanation, there was no explanation why he should have learning difficulties in any other way, there wasn't anything in the family...it wasn't like I mean I don't drink I don't smoke it's like you know all these other factors...I mean there was nothing that would ever contribute to that (David's mother, 2)

Emily's mother also did not report self-blame and said she did not dwell on what caused her daughter's difficulties.

I mean it has happened I mean it's something that you well I've accepted...we've not said oh you know was it somebody's fault or our fault or did something go wrong or whatever (Emily's mother, 2)

Like David's mother, she is confident that there was nothing to her knowledge that could have caused Emily's disability.

There was nothing really that indicated that Emily should have been anything other than normal in inverted commas her birth was natural...there was no emergency...it was all calm smooth sailing so there's nothing particularly in the background that I suppose sort of gives me an inkling (Emily's mother, 1)

There being 'nothing in the background' that seems likely to have caused Emily's disability renders the aetiology a mystery, which Emily's mother seems at peace with accepting.

For these mothers, although keen to know the cause of their child's disability and speculating about possible aetiology, self-blame was not reported to have been part of their thinking about cause. For most of the mothers, however, thinking about cause involved worrying they may be to blame for their child's disability. This was so even where there were strong suggestions that their child's disability had a genetic origin. Without certainty of aetiology, there is no reliable explanation for their child's disability. In the absence of a clear explanation, parents consider whether their own behaviour is culpable and mothers in particular seemed unable to fully eradicate the idea of self-blame. The cause of their child's difficulties remained a mystery. A discourse of risk and risk behaviours characterised mothers' discussion of what may have caused their child's disability. By referring to their conduct before, during and after pregnancy mothers' narratives reflected essential and idealised notions of mothering behaviours (Ryan and Runswick-Cole, 2008). The good mother in pregnancy was portrayed as one who ate well, avoided stress or doing too much,



and who didn't drink any alcohol, smoke, or take any medications. Making the right decisions in labour was also suggested as being associated with good mothering and, for one mother, making the right childcare choices in her child's early years. A discourse of conducting a good pregnancy was common to the narratives and risk was a significant organising principle in their thinking about cause.

In the dominant maternal discourse in Western culture, mothers are expected to play a vital role in improving their child's futurity, and may be held accountable for adverse outcomes and considered responsible for their child's disabilities (Fazil et al., 2002; Landsman, 2003; Landsman, 2009). They may endure stigma associated with atypically developing offspring (Fazil et al., 2002; Green 2003) and, while men and women contribute equal bio-material donations, women are assigned the responsibility for pregnancy outcome (Rapp, 1999, p88 cited in Landsman, 2009).

Landsman (2009) suggests that women are culturally held accountable for making the choices that lead to or preclude disabled children from being brought into the world. Maternal behaviour is monitored during pregnancy and maternal behaviour during, or even before, a pregnancy has wide currency throughout the world as a cause of a child's physical or cognitive impairment (Landsman, 2009). In Landsman's study of mothers of infants and toddlers with disabilities (Landsman, 1998, 2009), all of the women either struggled to determine what they might have done wrong to bring about a disability or felt they were being judged by others as having done something wrong during pregnancy. Women are expected to 'do everything right' in pregnancy to ensure the health of her baby, such as getting the right prenatal care; accepting diagnostic testing; eating healthy foods; and not smoking, drinking or using prescription or illegal drugs during pregnancy (Landsman, 2009, p17). On learning that their baby is disabled, most mothers at least initially consider that they personally may have done something to account for the disability. A feature of the narratives of the mothers in Landsman's study (2009) was mother's attempts to pinpoint the specific violation of the recommended prenatal behaviour to try to make sense of why their child was disabled. Landsman (2009) suggests that mothers reinterpret prenatal recommendations trying to locate a direct causal relationship between something they did during pregnancy and the pregnancy outcome.

Mothers in this way perform calculations of their level of culpability for what is culturally regarded as a dismal failure: a disabled child (Landsman, 2009, p23)

Women may search their memories sifting through what they had done or hadn't done that may have caused their child's disability; and it was the norm for women in her study to wonder about or even agonise over whether eating certain foods, working too hard, having an alcoholic drink before knowing they were pregnant, or using over the counter painkillers might have caused their child to have a defect. Echoing these findings by Landsman (2009), the mothers in my study searched through memories seeking behaviours or actions that may suggest culpability for their child's disability, looking both for what they had or hadn't done that may attribute for their child's difficulties. In this way mothers enact dominant cultural notions of the good and bad mother and self-blame was a common aspect of their thinking about cause. Even those mothers who did not express self-blame referred to ideals of good conduct in pregnancy and to avoiding risk behaviours associated with poor outcome. Indeed that their behaviour in pregnancy complied with these ideals was perceived as evidence that they could not be to blame for their child's disability.

#### *Fathers' self-blame*

All the fathers in the study also speculated about what may have caused their child's difficulties. Very few of the fathers, however, speculated significantly about self-blame and only one expressed feelings of guilt, although several fathers did speculate about whether they may have passed on a genetic disorder from their side of the family. Of the seven fathers who took part in the study, four speculated about their child's condition having a genetic origin, with two of these considering whether the problem lay with their side of the family. One father, Cameron's father, blamed himself for his son's disability and described intense long-term feelings of guilt. This was not seen in any of the other fathers' narratives.

As described in chapter 5 looking at parent narratives, Cameron's father felt to blame for his son's disability and the immense guilt that he felt about his son's disability caused him a lot

of distress. Even though he had been reassured that he wasn't to blame, he still felt that it must have been his fault. He described seeing doctors, psychiatrists, alternative therapy practitioners, taking medication and going to counselling in an effort to understand and resolve his feelings of guilt and his worry that he caused his son's disability, but with little success. The guilt was described as still as strong as it was when he first learned of his son's disability; 'it's not really any sort of better than it was before'. Unsure of whether Cameron's disability was genetic or not, he speculated about a variety of possibilities and described being tormented by 'what if' thinking.

There's not a specific point why I blame myself for it, it's a what if scenario you know, what if Cameron's like that because I had two or three beers the night he was conceived or something like that or what if you know there's something genetically wrong with me that made Cameron like that you know, it's because nobody can tell us why Cameron's like that, the mind starts working overtime (Cameron's father, 1)

In the absence of diagnosis, and facing unknown aetiology, he attributes the disability to his own risk behaviours or faulty genetic make up. He fills the void left by an absent diagnosis and aetiology with his own explanation.

People said to me...you know there's no way that it's been you, but you ask the question, what is it then? And they can't tell us so what are you supposed to do or think? (Cameron's father, 1)

He felt his wife did everything she could in pregnancy to make sure the baby was okay ('she did the best she could...I don't think she could have done anymore') so he rules out causes related to problems in pregnancy. In the absence of explanation, he constructs explanations based on self-blame: 'there's no other reason as far as I'm concerned' (1).

None of the other fathers expressed self-blame in this way for their child's difficulties, although all speculated about what may have caused their child's difficulties. Emily's father, for example, suspected that his daughter's condition was genetic ('I think it lies in the genes somewhere...I'm pretty much convinced', 1) and that it was likely to come from his side of the family.

Looking at my own side my mother had a brother who was quite severely handicapped and my sister's got a son who's quite severely mentally handicapped

and I'd say there's possibly some connection there...If I was being honest I probably think it's more likely to come from my side (Emily's father, 2)

Although he believed the genetic condition was likely to come from his side of the family, he was keen to avoid 'apportioning blame'.

Olivia's father described 'looking for answers' when it came to what caused his daughter's disability. He talked about being puzzled by there not being an answer and by not being able to 'explain why it is' (1). He reported feelings of frustration that 'there's no way of actually knowing what caused this' (1) and recalled going through a difficult time when his wife blamed herself for Olivia's difficulties. In seeking an answer, they had come across information about genetic conditions and had questioned which side of the family the disorder may have originated from. This was particularly problematic for one genetic syndrome that their daughter was tested for.

It can be traced back to the mother being missing certain chromosomes or the father producing two chromosomes and then it was kind of well what if it's you, what if it's me? (Olivia's father, 1)

Like Emily's father, Olivia's father said he was keen to avoid apportioning blame.

Abdul's father concluded that his son's difficulties were 'purely a random thing'. Although he considered his diet at the time of his son's conception ('I'm not sure how many portions of broccoli I should have eaten over that period of time in my life', 1) he largely shrugged off the idea that either himself or his wife may be to blame for his difficulties. He described, however, an earlier period of wondering whether the stress his wife experienced while pregnant may have had an impact on his son's health.

During the time [Abdul's mother] was pregnant she was going through a remarkably stressful time at work...I never you know said anything at the time...I just wondered at the back of my mind I wondered if that was part of it (Abdul's father, 1)

He described this speculation as 'purely a background thing' (1) that would occasionally pop into his head. Like most of the other fathers, he said he did not to focus on what caused his son's difficulties, and who might be 'to blame'.

At the end of the day it doesn't actually matter as far as I'm concerned...whether it was or not because you know that's just life kind of thing, you know, things happen (Abdul's father, 1)

Commonly, therefore, the fathers participating in the study speculated about what caused their child's difficulties, but tended not to apportion blame or blame themselves in the way that many of the mothers did. Little guilt was described regarding aetiology. The exception was Cameron's father who had experienced, and continued to experience, an intense amount of guilt and felt very much to blame for his son's difficulties.

In many ways, that all but one of the father's narratives was absent of self-blame and associated feelings of guilt, further augments the assertion that it may be women who are assigned the responsibility for pregnancy outcome (Rapp, 1999:88, cited in Landsman, 2009) and considered responsible for their child's disabilities (Fazil et al., 2002; Landsman, 2003; Landsman, 2009). If it is women who are culturally expected to 'do everything right' (Landsman, 2009, p30) then it is not surprising that fathers' accounts commonly do not feature self-blame. Cameron's father was the only father to express self-blame and of all parents, both mothers and fathers, seemed the most tortured by feelings of self-blame. Yet his blaming of himself partly relied on notions of good mothering and idealised conduct in pregnancy; as his wife had done everything right in pregnancy it made sense to him that he must be to blame. A small number of fathers made reference to the cultural expectations of how a mother should behave in pregnancy. Moy's father, for example, referred to his wife's extreme stress in pregnancy, citing this as one of the reasons they should not go ahead and have another child. Abdul's father worried that his wife had worked too hard and been too stressed. He said he hadn't dwelled on this concern, and described it as being put 'to the back of his mind'. That there were more mothers than fathers in the study makes it difficult to comment on the implications of this finding, beyond highlighting that almost all mothers (16 of 19) and only 1 (of 7) father expressed self-blame. This resonates with relevant literature on cultural expectations of mothers' conduct in pregnancy and their responsibility for pregnancy outcome.

### 7.3 Risk of Recurrence

Many parents reported that not having a diagnosis had an impact on their decision about whether to have more children. Fourteen parents said that not having a diagnosis had impacted on their lives in this way; with 11 parents saying that they decided not to have another child due to not having a diagnosis (for 2 of these parents this decision was ongoing), and 3 parents reporting that they significantly delayed having another child due to not having a diagnosis. Unknown risk of recurrence was commonly the reported reason for delaying having another child or deciding not to have another child. Parents could not be sure whether their child's condition was likely to recur as the precise genetic cause was not known and therefore the risk of recurrence could not be assessed.

#### *Impact on Future Reproductive Choices*

The majority of the fourteen parents who said not having a diagnosis had affected subsequent reproductive choices had decided not to have another child.

We also decided not to have any more children as we didn't know if Anna's condition was genetic and further children would be affected (Anna's mother, IF, USA)

Three parents (with an additional two parents being undecided at the time of interview) had decided to go ahead and have another child but had delayed this decision due to not having a diagnosis. For the remaining parents (10) this issue was not relevant as they had not planned on having any more children regardless of having an undiagnosed disabled child.

Of those parents who decided not to have another child, the decision process was often described as lengthy and difficult. Cameron's parents described this process of considering having another child, then deciding against it due to the unknown risk of recurrence. Both parents in their interviews talked about how they had hoped for more than one child. Cameron's mother in particular spoke of her hopes for a large family prior to the birth of her

son. After his birth, however, they found it difficult to make a decision about whether to go ahead and have another child.

The paediatrician once said to us that they couldn't guarantee that if we did have another child that the child would be the same as Cameron (Cameron's mother, 1)

They delayed their decision about having another child, worried about the risk of recurrence. Cameron's mother spoke with sadness about how she was now, at the time of the interviews, too old to have another child. Cameron's father also described a process of being unable to 'commit to having another child'. He said:

We couldnae be guaranteed that we wouldn't have another child with or without problems so it swayed us towards not having because they couldn't guarantee us...I'm sure at the time they said the chances are you would have a healthy child but it didn't overcome the doubt in our mind (Cameron's father, 1)

Not having a diagnosis was not always the only factor informing the decision of whether to have another child. Some parents reported that not having a diagnosis, along with the increased care needs of their disabled child, contributed to their decision. For Cameron's parents, although not having a diagnosis was a very significant factor in their decision, that Cameron needed 'attention one hundred per cent of the time' (Cameron's father) was also a factor. Similarly, Eva's mother said that not having a diagnosis alongside Eva's care needs were factors in deciding not to have another child.

We decided to have no more children, partly because of the hard work looking after Eva, but I wouldn't have done anyway since I could not bear for this to happen again (Eva's mother, IF, ENG)

Moy's parents had also decided not to have another child. Their daughter was an only child, with both parents keen to expand their family. They had consulted with the geneticist about the risk of recurrence, but without a diagnosis they could only speculate on the risk of having another child with the same condition. They had decided not to take the risk, fearful that their second child would need as much (or more) care than their daughter. As the genetic risk could not be known, they were afraid that 'lightening could strike the same place twice' (Moy's father, IF, USA). As Moy's mother explained, there was no test that could give them an idea of the risks involved in having another child. She had been looking

for reassurance that a second baby 'would be fine'. Without this, they felt they could not go ahead and have another child.

I wanted to find out what the chances are that this could happen again well the fact that we don't have a diagnosis you know I don't think there's anything that they can test us for if I knew the baby would be fine I would have another in a heartbeat (Moy's mother, TI, USA)

Two parents (Olivia's parents) were undecided about having another child at the time of the interview study. Not having a diagnosis was reported by Olivia's parents as having a significant impact on their decision about future reproduction and they had decided not to try for another child until they had a clearer idea of their daughter's diagnosis.

Three parents had decided to go ahead and have another child but had delayed this decision due to not having a diagnosis. For two of these parents, Caroline's mother and Abdul's mother, the decision to go ahead and have another child was grounded in 'gut feeling' (Abdul's mother) and 'optimism' (Caroline's mother) rather than scientific knowledge or proven statistical risk.

For Caroline's mother, not having a diagnosis had a 'huge impact on deciding to have a third child, an enormous impact yes we definitely delayed that' (1). She had been pregnant already when she realised that her eldest child, Caroline, had significant development difficulties. It was the decision to have a third child that was impacted by not having a diagnosis. Caroline's mother said it was 'optimism' and 'just really wanting another' that in the end helped them to make their decision to go ahead and have another child, although she also referred to probability; 'probably deciding that on balance the chances were still greater of having a healthy child compared to not.' (1) This was a difficult decision, particularly considering that her first-born child (born before Caroline) had died soon after he was born. Although she referred to probability, she saw estimations of probability as based largely on 'guess work' in the absence of diagnosis. The decision to go ahead was described as an emotional instinctive one, rather than being evidence based, as she described deciding 'to take the chance' (1). As well as the implications of not having a diagnosis, the decision to delay having another child was also attributed to the physical demands of



looking after their other two children and the additional demands of looking after a child who needed extra care.

Similarly, Abdul's mother described deciding to take the chance ('it was something that almost we just came to by instinct rather than be rational about it' 1) by going ahead and having another child. She described how initially she sought to make a decision 'on the basis of knowledge', seeking from consultants 'an indication of the risks for a second child' (1). They could not find any certainties about the likelihood of recurrence and, although the geneticist gave them a probability of recurrence, this was subject to change over time. Abdul's mother described how she came to the decision to have another child by 'gut instinct' rather than making a 'rational' decision.

Eventually it was something that almost we just came to by instinct rather than being rational about it, because there was a long time we couldn't even go there...I think I have to do everything I mean you know we sort of talk about it and say well here's the risks...you know you think no matter what we do whether it's a child with problems or a child without it's going to be good for him to have a sibling...all these things are going on in the back of your head but we still couldn't quite actually take the decision and then eventually my hormones just went yeah go for it! (Abdul's mother, 1)

This decision was described as 'daunting' at the time of our first interview, as it was not known yet whether their second child would have the same disability. Abdul's father also described this process of deciding to go ahead and have another child. In his account of the decision making process, the geneticist reducing the estimated risk of recurrence from '1 in 4' to '1 in 10' was said to inform his decision, along with the positive attitude of their keyworker.

By the time of the second interviews with Abdul's mother and father their daughter was born. She had normal development with no apparent difficulties. Abdul's father said of their decision: 'I think we just decided to have a go and see what happened and fortunately things have worked out okay' (2). Although not having a diagnosis delayed the decision to have another child for this family, they still decided to go ahead with having more children, taking a chance when faced with uncertainty of aetiology and risk of recurrence.

In this section, I have reported that not having a diagnosis had an impact on future reproductive choices for many families as they were unable to assess the risk of recurrence. Fourteen parents reported an impact on future family planning, with 11 parents reporting they had decided not to have another child. Certainty of aetiology relies on certainty of diagnosis. Although genetic disorders are not necessarily inherited, and may occur *de novo*, without a specific diagnosis it is not possible to rule out an inherited genetic disorder and parents of undiagnosed children face difficult reproductive choices that must be made without knowing specific recurrence risks.

As suggested above, Landsman (1998, 2009) proposes that women are culturally held accountable for making the choices that lead to or preclude disabled children from being born. She asserts that reproductive technologies, including pre-natal testing, put pressure on mothers to produce perfection and reject the 'abnormal'. Notions of attribution and personal responsibility for the birth of a disabled child may have increased due to prenatal testing and the resultant attitude that disability is something that can be screened for and, ultimately, eradicated (Marteau and Drake, 1995). Ginsburg and Rapp (1991) suggest that technology becomes the resource that pledges to produce the perfect child; and women may be increasingly held morally accountable for a pregnancy's outcome.

Reproductive decision making for parents of existing children with genetic conditions is complex. Parents can be in a unique position with regard to choices and dilemmas posed by prenatal screening and testing options offered to parents at risk of having children with genetic conditions. Kelly (2009) suggests that parents in this position often choose not to choose: avoiding future pregnancies, declining prenatal testing for subsequent pregnancies, or limiting testing to 'for information only'. By choosing not to choose they negotiate the difficult decisions associated with the options presented by reproductive technologies and the ambivalence represented by the desire to have another child and the emotional impact of feeling they may be choosing not to have another child like their own.

Several studies suggest that for most people, genetic responsibility is enacted by not passing the gene to their children either by using genetic testing to select for an unaffected embryo, or by choosing not to have (more) children (Hallowell, 1999; Kelly, 2009). Women identified as carrying, or potentially carrying, a gene causing disability may have to reconcile desire to

have more children with a sense of oneself as genetically at risk (Raspberry and Skinner, 2011). In much of the literature looking at genetic responsibility and citizenship, the genetically responsible citizen is portrayed as one who makes use of genetic information to direct her own and family's health (Kerr and Cunningham-Burley, 2000). A sense of genetic responsibility may be particularly pronounced when families are making reproductive decisions, when the genetically at-risk individual confronts the possibility of passing on the defective gene (Parsons and Atkinson, 1992). However, making reproductive decisions in light of already having a disabled child involves beliefs about personhood and societal acceptance of children with disabilities, and deep ambivalence towards actions that may reflect choosing against a disabled child (Kelly, 2009).

For parents in my own study, reproductive decision-making was affected by absent diagnosis. With lack of clarity about whether the condition was inherited, and no possibility for pre-natal testing, parents had limited options available to them. They could either choose not to have any more children, which many did, or they could go ahead and have another child uncertain of whether that child would also be affected, which a small number of parents did. The 3 parents who choose to have another child described delaying this difficult decision, their decision resting on feelings of 'optimism' (Caroline's mother) and 'gut feeling' (Abdul's mother) rather than scientific evidence of recurrence risk. Both families went on to have children who were not affected by the disability.

## **7.4 Conclusion**

This chapter has considered how parents perceived the absence of diagnosis impacted on issues related to knowing the cause of their child's disability. Two themes were identified: self-blame, and impact on assessing the risk of recurrence. All of the parents in the study speculated about what may have caused their child's difficulties. Most mothers (16 of 19), and one father, reported that they speculated about whether they were to blame for their child's disability, and around half of mothers (9) and one father expressed feelings of guilt related to aetiology. Many parents (14) reported that not having a diagnosis had an impact on their decision to have more children; with 11 parents reporting deciding against having more children due to not having a diagnosis (for two of these parents this decision was on-

going), and three parents reporting that they significantly delayed having another child due to not having a diagnosis. For the remaining parents, the decision was not relevant, primarily because they had not planned further children in any event.

These findings resonate with the findings from other research work with families that identify cause as an important aspect of having a diagnosis for parents (Rosenthal, Biesecker and Biesecker, 2001; Lewis, Skirton and Jones, 2010; Watson, 2008). Parental desire for diagnosis is linked to a need to assign blame for their child's anomalies (Rosenthal, Biesecker and Biesecker, 2001) and, in the absence of diagnosis, parents are concerned about recurrence risks in future pregnancies and for other family members (Rosenthal, Biesecker and Biesecker, 2001; Lewis, Skirton and Jones, 2010). In the study by Rosenthal, Biesecker and Biesecker (2001) several families had postponed or foregone having more children due to uncertainty about recurrence and parents who did go ahead and have more children reported anxiety during pregnancy, with two families in their study subsequently giving birth to a second child with anomalies. Parents of children with unidentified anomalies might be more susceptible to self-blame and recognising a cause may help reduce feelings of guilt or parental blame as parents commonly wonder if they have done something to cause their child's difficulties (Rosenthal, Biesecker and Biesecker, 2001; Watson 2008). Burden (1991) postulates that not having an understandable cause for a disability is one of the reasons parents may experience guilt in reaction to having a child with a disability. Burden suggests that when the etiology of the disability can be satisfactorily explained, there will be a reduction in feelings of guilt, but as long as the etiology remains 'shrouded in mystery' the likelihood of self-blame is increased (Burden, 1991, p 333).

The search for cause is embedded in our culture, intimately linked with the constructs of responsibility and blame. Parents of children with severe, chronic diseases may commonly experience an intensely felt need to find an explanation or cause for their child's disorder. Guilt and blame are reported as common responses to a child's diagnosis (Kessler S, Kessler H, and Ward P, 1984) and parents make causal attributions (including self-blaming attributions) for their child's illness as a defense against strong feelings of helplessness and meaninglessness (Weil, 2000). Parents may be more likely to blame themselves for their child's condition when there is no clear answer regarding the cause of their child's difficulties (Nixon and Singer, 1993). Avdi et al., (2000) report that it is particularly common

for mothers to situate the cause of their child's difficulties with themselves, their own behaviour or events during the time of birth or during pregnancy.

Referring to risk behaviours perceived to be associated with poor outcomes in pregnancy was common to most parent narratives, either when expressing concerns that they may have caused their child's disability or appealing to a logic that they couldn't have been to blame as they followed recommended advice. As a feature of later modernity, risk is an organising principle at both the institutional and individual level (Hallowell et al., 2006). Risk, a principle at the hub of reflexive modernity (Beck, 1992; Giddens, 1990, 1991), is now a significant preoccupation for lay people and experts. Assessing risk has become a key element of public health, with discourses of risk concentrating on risks at the environmental level, as well as risks associated with lifestyle choices made by individuals. With the proliferation of discourses of risk as their cultural backdrop, it is understandable that parents referred to risks at the environmental level and risks associated with lifestyle choices when trying to account for their child's aetiology. Thus they speculated on, for example, exposure to radiation at work; stress caused by work or life events; toxins in pregnancy including alcohol and chemicals found in non-organic food; risks during labour; and genetic events, both random and hereditary. Even when a child's disorder is caused by genetic mutations out of their personal control, parents can still experience feelings of guilt associated with passing on the condition (Hallowell et al., 2006).

This analysis of the parents' account suggests that, in the absence of diagnosis, deviance (Goffman, 1959) associated with responsibility for the onset of their child's disability may never be abrogated, particularly for mothers who culturally are held to carry more of the responsibility for pregnancy outcome. Without a clear diagnosis, there are no boundaries to individualised blame for adverse outcomes. Actions or omissions of the parent (like having a glass of wine in pregnancy or not eating organic food) cannot be ruled out and the parent cannot prove or disprove their culpability. The questions parents ask about what may have caused their child's disability ('Did I do something wrong in pregnancy?' 'What have I done that I damaged my chromosomes?') cannot be definitively answered. Without a diagnosis, the question of whether they may in some way be responsible for their child's disability can not be put to rest.

This chapter has considered how absent diagnosis had an impact on how parents conceived of the temporal realm of the past, and how that featured in decision making about the future

with reference to future reproductive decision making. In the next chapter, chapter 8, I consider how absent diagnosis impacted on how parents thought about the future by looking at their perceptions of uncertain prognosis in the absence of diagnosis.



## **Chapter 8      Living with Prognostic Uncertainty**

### **8.1      Introduction**

### **8.2      Parents concerns about their child's prognosis**

*Health, development and education*

*Adulthood and life expectancy*

### **8.3      Ambivalent feelings towards uncertain prognosis**

### **8.4      Strategies for dealing for uncertain prognosis**

### **8.5      Conclusion**



## 8.1 Introduction

In chapter 7, I considered how not having a diagnosis had an impact on the way parents looked back in time to what caused their child's difficulties and how not knowing what caused their child's difficulties in turn had an impact on the future, in terms of making reproductive choices. In this chapter the temporal domain of the future is further considered, as I examine whether parents perceived absent diagnosis had an impact on how they thought about their child's prognosis. All parents talked about the uncertainty of their child's health or development. A high number of parents (19) related this uncertainty to absent diagnosis. Twelve parents spontaneously related absent diagnosis to not knowing their child's prognosis when telling me their story at the beginning of the interview or responding to my general question about whether not having a diagnosis had an impact on their lives. The remaining parents raised issues relating to prognosis when asked whether they thought not having a diagnosis had an impact on how they thought about their child's prognosis. Only a very small number of parents (2) felt that not having a diagnosis had not had an impact on how they perceived their child's prognosis or the future, although several parents (5) were unsure whether uncertainty of prognosis was related to non-diagnosis or more generally to the issues of not knowing what the future holds for any disabled child.

Parents reported not knowing what to expect in their child's long-term development, as well as in the more immediate future. They worried about their child's development, health, education, adult life and, in a small number of cases, life expectancy. Commonly, in the absence of diagnosis, they did not know what to expect and they found it difficult to plan for the future. This chapter will look first at parents' concerns about their child's prognosis (8.2) including concerns about health, development and education, and concerns about adulthood and life expectancy. Ambivalent feelings towards uncertain prognosis expressed by parents will then be explored (8.3), followed by strategies used by parents for dealing for uncertain prognosis (8.4).

## 8.2 Parents concerns about their child's future in the absence of diagnosis

All parents spontaneously referred to the uncertainty of their child's prognosis or not knowing what to expect for their child or family in the future

I've no sense of what will happen for our family and how to prepare for that (Jessica's mother, IF, AUS).

You don't know what's going to happen in the future, you don't know how your child's going to develop (Caroline's mother, 1)

I fear for the future (Eva's mother, TI, ENG)

Nineteen parents specifically related absent diagnosis to uncertainties about the future, reporting that not having a diagnosis made it more difficult for them to know their child's prognosis or what to expect in the future.

With not having a diagnosis, I don't know what to expect from his future (Ismail's mother, 1)

For some parents, dealing with an uncertain future was their main worry. Emily's father, for example, said that the 'main concern' they had was 'for the future really, it's what is going to happen to her in the future because we don't know whether she's going to get much better...we see it as uncertain, there's no doubt about that'. Calum's father described his son's unknown prognosis as a 'massive' issue (1). As he worked with vulnerable adults he was aware of some of the issues they faced and was uncertain about whether Calum would be vulnerable as an adult.

Several parents said they recognised that the future was uncertain for everyone, but was particularly so without a diagnosis when there may be increased uncertainty of what to expect in the future. Uncertainty in the absence of diagnosis was seen as a matter of degree. When referring to named diagnoses, some parents talked about having guidelines and parameters about future expectations that were absent without diagnosis.

Nobody knows exactly but it's still helpful to have guideposts and I think there are more if you have a diagnosis then at least you've got more guideposts that you're

heading toward (...) there are no benchmarks there, you know, you don't have any sort of other benchmarks with other folks (Emily's mother, 1)

When you've got the diagnosis...you're going to have a fair idea of what to expect...you're going to get milestones basically like a normal child but obviously with different milestones whereas if you're trying to cope with a child without diagnosis you don't have these milestones (Calum's father, 1)

Not being able to make plans for the future was suggested by some parents as being particularly problematic. Parents wanted to be able to make plans for the future and to know what kind of a future their child might have so they could prepare themselves.

At least if we know what it is we'll be able to make plans to face it (Calum's mother 2)

We're still in limbo and not having a diagnosis...you can't make sense of things, you can't plan for the future because you don't know what the future's going to hold (Jessica's mother TI)

The big one is being able to look ahead to plan, we can't look ahead (Calum's father 1)

One way in which absent diagnosis was perceived to impede being able to access knowledge about prognosis was that parents reported that they were keen to seek prognostic information by making contact with other families, which wasn't possible without a diagnosis. They could not, therefore, look to other families with children with the same condition to see how their child had developed. Parents were particularly keen to be in touch with families with a child with the same condition who was older than their own child. Having a diagnosis was perceived as allowing comparison across time in that parents could potentially gain insights by making contact with families with older children.

If the doctors came back and said Olivia has this or this then there could be another there must be someone else out there who's went through all the generations that Olivia's going to go through in her life and to give you an idea of how they coped and what happened to them through life, not saying that's the way Olivia's path would follow because I know everyone's different and everyone's unique, but it would still give you that idea (Olivia's mother, 1)

[with a diagnosis we could] speak to someone that would be able to tell us when Calum is 15 and when Calum is 18 you can expect x, y and z (Calum's father, 1)

Parents were keen to learn from others who had been through the same experiences and the value of relating experience between families was recognised.

We thought that if we could find out what was wrong then we could maybe speak to people in similar situations and try and get some insight you know, their girl went through a really frustrating period or their boy and how they best dealt with it...because I think you can learn from other people's experiences that have went through similar situations to yourself (...) I mean everything's in comparison to something else isn't it, so you've nothing to compare it with (Olivia's father, 1)

In addition to a generalised concern about an uncertain future, parents also described specific concerns regarding their child's development and future, both in the short and the longer term.

#### *Health, development and education*

Parents' concerns about the future in the absence of diagnosis included not knowing what to expect in terms of their health or development, and feeling less able to help their child by providing the right treatment, education, or therapy.

Parents expressed concerns about health issues in the absence of diagnosis. More common childhood disorders, like Down syndrome for example, often come with associated health issues that can be screened for or monitored. Even relatively rare conditions, like rare myopathies (muscle disorders), are often known to come with particular health issues like respiratory or cardiology problems. Without a diagnosis, however, it is difficult to predict health issues or to be aware of the range of possibilities so that a child can be monitored in case they arise. One parent in particular, Abdul's mother, described a very difficult time when Abdul experienced a variety of health issues across a short period of time. In the absence of knowing what to expect, Abdul's mother feared what she was going to find out next. She described this period of ill health as frightening as she felt unable to anticipate the problems he might have. As each new difficulty emerged, she felt anxious not knowing what was coming next; 'it was just always living with what's coming next and not knowing' (1). She related this to absent diagnosis.

A number of parents referred to the impact of absent diagnosis on not knowing what they should expect from their children in terms of development and found it difficult to optimise their therapy or treatment without adequate knowledge about their expected development. Moy's parents felt absent diagnosis made it more difficult to plan treatment and therapy. They worried that they did not know where their daughter might 'plateau' in her development. Moy's father felt that if she had a diagnosis her treatment could be 'based on other similar patients' and she could be helped to follow a more 'predetermined course of action.' Peter's mother felt that a diagnosis would be accompanied by a better idea of how to help her son; without a diagnosis they lived with the fear that they weren't doing enough to help him as they didn't know what to expect from him and what the future would hold for him. Claire's mother felt that, without a diagnosis, she couldn't know what her daughter would be capable of. She regretted not being able to piece together all the information to create a clearer idea of what to expect from her daughter's development.

If they gave us a word we'd be able to look it up each different bit, we'd be able to put it together and know what she'll be capable of (Claire's mother, 2)

Ismail's mother said that with lack of diagnosis it was difficult to ensure he was getting the right treatment as she didn't know what to expect from his development.

I'm never sure you know like maybe I'm pushing him too much or not pushing him enough em I don't know what his abilities are you know, what he can really achieve...because of lack of diagnosis it didn't really prepare you for what extent his difficulties were (Ismail's mother, 1)

She worried she had not made the right choices for him, particularly with reference to his school placement. For Abdul's mother lack of diagnosis contributed to her feeling that his developmental future was unknown ('the lack of diagnosis means you can't say well this is like what he's going to be able to do...we don't know I mean he might stop developing completely' 1) and Jessica's mother also linked uncertainty regarding her daughter's future development with inadequacy of treatment and therapy plan.

If we had a diagnosis we would have a better handle on what it is that we should do for her. What are the most important things for us to target. What type of therapy is most likely to be successful (Jessica's mother, IF, AUS)

In thinking about prognosis, concerns were also raised about school placement and difficulties making decisions about the right choice of school in the absence of diagnosis. This was a particularly timely consideration for David's mother at the time of our first interview as she was making choices about school placement for David. She worried about his future education and what would happen when he progressed from nursery to school. Not knowing what to expect from his development was also having an impact on her decision to buy a family home, as she wanted to make sure she considered his educational needs when looking at where they might relocate. As she didn't know what his educational needs might be, this was difficult.

It's like I don't really know for certain how he's going to perform in school and I don't really know how he's going to succeed in life (...) Shall we stay in this area? Is the school in this area any good? Will they give me additional support needs? Or will I put him in this school in this area or should I go and look at houses in this area? My concerns are actually not just looking at the short term needs but the long term needs as well, as a family where do we go? Where do we stay? I can't foresee what's going to happen when he's 5 or 7 or whatever it's em the diagnosis you know (David's mother, 1)

Absent diagnosis was seen by these parents to impact on knowing what to expect from their child's future health and development. This had real effects in terms of worry and also choices, both choices they faced at the time and anticipated choices in the future. They expressed concerns about making the right decisions for their child, in terms of treatment, education and therapy. Concern about anticipating health issues was also expressed. Some parents may perceive a diagnosis as offering parameters around what to expect in the future in terms of their child's health or development. In the absence of a diagnosis, they are required to make decisions for their child about therapy, education and treatment without prognostic information. It seemed that absent diagnosis had an impact on how confident they were in their own decision-making. These aspects of an uncertain future were seen to be tied to, or exacerbated by, lack of diagnosis.

Many parents perceived that without a clear diagnosis it was difficult to access information about their child's future or how they may develop over time. A number of parents considered the long-term implications of this uncertainty. In this context, concerns were expressed about their child's potential for independence as adults, whether parents would be life-long carers, who would care for their child in their absence, and not being able to protect their child as easily when they reached adulthood. Although these concerns may be relevant for all parents of disabled children, here I report parent perceptions that absence of diagnosis had an impact on thinking about the long-term future. Parents raised these issues of their child's future adult life when talking about how not having a diagnosis had an impact on their lives, therefore they directly related absent diagnosis to their concerns about the long-term future. The most commonly expressed worry in this context was whether their child would be independent as an adult.

The main concern we have is for the future really...what sort of level is she going to get to? Will she even be able to cope independently? (Emily's father, 1)

We also fear for the future as we don't know if he will ever be independent from us (Peter's mother, IF, AUS)

We've asked the question what is his end goal likely to be?...you can't think ahead, really for his future what level of independence is he going to have (Abdul's mother, 1)

Parents' concern about their child's future independence was sometimes accompanied with related concern about who would care for their child in adulthood.

What's going to happen to Cameron in terms of you know when he leaves school? Is he going to be able to live himself?... Are we going to sort of provide for him for the rest of his life? (Cameron's mother, 1)

Some parents anticipated that they may be life-long carers for their son or daughter and expressed worry about what would happen to their child when they were no longer able to look after them. Two parents expressed concerns that the caring task would fall to their other children. Abduls' father mentioned that his daughter would have her own life to think

about and may not be able to care for her brother. Emily's father had similar worries and didn't want his elder daughter to feel that Emily was her responsibility.

Eva's mother had a general concern about not being able to protect Eva, in the way she currently did, once she was in adulthood.

I worry about you know at the moment she's a child and I can treat her like a child and protect her like a mother protects a child, but when she gets older, when she's an adult...I've no idea what she's going to be like and I'm concerned about the fact that I'm not going to be able to protect her like a child (Eva's mother, TI, ENG)

A small number of parents (Peter's mother, Claire's mother and Andrew and Lee's mother) worried that their child's life span was unknown and made comments about not knowing how long their child might live; a concern that was attributed to not having a diagnosis.

They cannae give us a life span on her, they could say oh she's got one year left, she's got ten years left, we just don't know and that does make it hard. At least when you've got a diagnosis you could look it up and think oh well that person there's lived to that age and you could have a bit of an idea...that could be the hard bit, not knowing (Claire's mother, 1)

One parent expressed concern about not being able to rule out with certainty that their child had a degenerative or progressive disorder. In the absence of diagnosis, they worried about the stability of their child's condition.

We are very unclear as to what the future holds. I still worry that it may turn out to be a degenerative condition and he will die young (Peter's mother, IF, AUS)

These parents raised concerns about the future in terms of their child's long-term development and adult life. This section has looked at parents' concerns about their child's prognosis and suggested that many parents perceived the future to be more unknown in the absence of diagnosis. While many parents (19) expressed concerns about their child's future and linked these concerns to not having a diagnosis, around a third of parents held an ambivalent position to 'not knowing'.



### 8.3 Ambivalent feelings towards uncertain prognosis

Eight parents described ambivalent feelings towards knowing their child's prognosis or recognised that there was a positive element to not knowing what to expect in the future. This ambivalence seemed to rest on comparing the situation of not having a diagnosis to having a diagnosis that came with a poor prognosis. In this eventuality, it was possible that not having a diagnosis was more beneficial. Ignorance was 'bliss', if the alternative was a poor outcome.

It's still quite a lot to cope with [having a diagnosis of Down's syndrome] isn't it? Then they know what's ahead of them, maybe that's worse sometimes, I don't know, ignorance is bliss (Calum's mother, 2)

Eva's mother expressed ambivalence about unknown prognosis and appeared to feel comforted by not having to deal with certainty of prognosis.

I have mixed feelings on prognosis. In some ways I really want to know how she will be when she grows up, whether she is likely to progress or just stop...but sometimes it can be a comfort not to have to deal with certainties (Eva's mother, IF, ENG)

She acknowledged that even for children with normal development, the future was unknown to a certain extent. Without a diagnosis, she felt the possibilities for her daughter's future were more open; 'sometimes having no diagnosis actually allows you to believe that things aren't final'. However, when prompted to talk about this further, she talked about fearing getting a diagnosis with a poor prognosis, and felt that not having a diagnosis would be preferential to having a diagnosis she didn't want. She retained hope, saying: 'it could be worse, they could be telling me things that could be worse, more final somehow'. She described feeling afraid at the times they seemed close to getting a diagnosis; 'every time I think we may be coming close I actually find it quite scary because while there isn't a diagnosis at least I can think to myself well at least she hasn't got this and hasn't got that' (all extracts from TI). Her ambivalence at these times rests on comparing the situation of not having a diagnosis with having a diagnosis with poor prognosis.

Similarly, Peter's mother was glad that they didn't have a diagnosis of 'something horrible and life-threatening' (TI, AUS). Moy's father described having a diagnosis as potentially being 'a double edged sword' (IF, USA) when it came to prognosis, in that it could offer the possibility of a more certain prognosis, but that this might bring with it a worrying prognosis. For Moy's mother, not having a diagnosis left open the possibilities that her daughter may talk or walk, and she recognised that with a diagnosis often came a continuum of possibilities for prognosis, including a worst-case scenario. This left her feeling ambivalent towards knowing the prognosis.

Not having a diagnosis is an okay thing because I think well maybe there's a chance that she'll maybe talk or maybe she'll walk or if she because you don't know and with a diagnosis well whatever diagnosis it would be though there's always the worst case scenarios of that and just like a continuum of better or worse so I don't know (Moy's mother, TI, USA)

Caroline's mother had experienced considering a diagnosis with a very poor prognosis for her daughter. Initially, Caroline was suspected to have a condition that was associated with significant health problems. Caroline's mother felt 'better off' without a diagnosis when this condition was ruled out; 'there were so many negative things associated with that diagnosis that after that I maybe thought well maybe I'm better not knowing' (1). In this way, not having a diagnosis was better than having the named diagnosis originally suspected, although not necessarily better than having a different diagnosis.

Around a third of parents described ambivalent feelings towards knowing their child's prognosis or recognised that there was a positive element to not knowing what to expect in the future. Commonly, this ambivalence rested on the idea of having a diagnosis with a very poor prognosis, which was seen as potentially worse than having no prognosis.

#### **8.4 Strategies used by parents for dealing for uncertain prognosis**

Eight parents spontaneously described strategies for dealing with the uncertainty of the future. Parents were not asked directly about how they coped with uncertain prognosis or about strategies to lessen the impact of prognostic uncertainty. The strategy most commonly

described by these parents for dealing with uncertain prognosis was managing time by breaking it into smaller units and focusing more on the present than the future.

I just take each day at a time (Ismail's mother, 1)

We deal with the immediate future and enjoy the here and now....I take each day as it comes (Caroline's mother, 1)

Abdul's mother described how she coped with an uncertain future by breaking down each stage of her son's life into smaller steps. She felt that if she dealt with each small step along the way ('we just take it step by step', 2), then by the time she got to the later stages of his development she would be ready for whatever she faced. Having gone through a period of intense worrying about his health and what to expect next from his development, she felt that she had had to 'train' herself not to think very far ahead. She related having to adopt this strategy for dealing with an uncertain future to not having a diagnosis.

The only way I think I'm going to be able to cope with the future is that by the time it comes I will you know I've coped to here and I just think we're with him every stage... I can't conceive what it's going to be like...the only way I can sort of manage and cope is just to think well by the time I get there I'll have done all the steps to get there...we'll just see one step at a time...that's the thing again because of his lack of diagnosis (Abdul's mother, 1)

Abdul's mother had adopted a strategy of breaking down time into more manageable units, taking one step at a time, in order to cope with the uncertainties of the future. Both Abdul's parents felt this 'wait and see' approach worked for them when trying to deal with the uncertainties of the future. His father said 'we'll cross those bridges when we come to them' (1) when talking about his son's future life. He felt he didn't worry about what the future holds as he would deal with each situation as it arose. Practically, he was planning ahead for Abdul by saving his Disability Living Allowance for him so that it might help him in the future. He felt that this was all they could do for him, in the absence of knowing what to expect from his son's development and future life; 'that's probably all you know I think we can realistically do...those things, but there's no grand plan, no timeline, no targets or anything like that' (1). They managed their expectations by not thinking too far ahead, having 'no grand plan'.

Calum's mother also described breaking down time into smaller units to manage uncertainty, taking things as they happened and not making plans.

We just take each day as it comes...we just have to take it you know as it happens...we just get on with it and see what happens (Calum's mother, 2)

Calum's father also dealt with uncertainty by limiting his temporal focus, looking to the present rather than the future and tackling issues as they arose, 'crossing those bridges when we come to them'.

This strategy of focusing on the present was seen across 8 parent narratives. The negative impact of thinking ahead to the future was recalled by several parents, who reported that they had experienced significant anxiety when previously contemplating the future for their child. Jessica's mother, for example, said she had 'learned to live with' uncertainty and accept that the future was unknowable. This move towards acceptance followed a long period of depression and anxiety when she felt she 'couldn't make sense of it' (TI, AUS). Her strategy of trying not to think too far ahead had helped her fears for her daughter's future.

The strategies used by parents to help them deal with an uncertain future involved breaking down time into smaller units, not thinking too far ahead and instead taking each day or stage in their child's life at a time. The impact of uncertain prognosis was minimised by a focus on the present, rather than thinking ahead which could cause worry or anxiety.

## 8.5 Conclusion

All parents referred to the future or their child's prognosis as uncertain, with 19 parents specifically linking this uncertainty of prognosis and what to expect in the future to not having a diagnosis. Parents expressed worries about their child's development, health, education, adulthood and life-expectancy. In the absence of diagnosis they found it difficult to plan for the future. The uncertainty regarding their child's future was commonly felt to be uniquely unknown, with absent diagnosis seen to be tied to or exacerbating uncertainty about the future. We all live with uncertainty, but for parents of disabled children without aetiological or prognostic certainty, the world may seem even more unpredictable.

These findings resonate with the findings from other research work with families that identify uncertainty of prognosis as an important aspect of living without a diagnosis (Rosenthal, Biesecker and Biesecker, 2001; Lewis, Skirton and Jones, 2010; Watson, 2008). In other research work with families with disabled children it is reported that one of the most common reasons given for wanting a diagnosis is 'to know about the future' (Lewis, Skirton and Jones, 2010) and gain knowledge about future expectations (Watson, 2008), and this was reported as being the motivation cited most often by parents seeking a diagnosis (Rosenthal, Biesecker and Biesecker, 2001). Diagnosis can be seen by parents as an antidote to uncertainty, a label that can transform the problem from something poorly understood to something knowable with a predictable future (Avdi et al., 2000), and parents want information to guide expectations and plan for their child's future (Makela et al., 2009). Parents who experience greater uncertainty feel less control over their child's medical condition which may lead to less effective coping and poorer adaptation (Madeo et al., 2012).

Other research has established that parents desire a diagnosis that will give an insight into their child's future and seek prognostic information about: future threats to their child's health; the boundaries of their child's capabilities so they could formulate realistic expectations and goals; and how much progress their child will make in overcoming his or her disability (Rosenthal, Biesecker and Biesecker, 2001). In other work with families looking at the significance of diagnosis, parents were keen to predict their child's future development and their future obligations, including what care their child would need as an adult and whether they would need to live at home (Rosenthal, Biesecker and Biesecker,

2001); whether they would have to care for their child for the rest of their lives (Lewis, Skirton and Jones, 2010); and who would care for their child when the parent was no longer there to provide care (Madeo et al., 2012). Parents had concerns about their child's medical vulnerabilities and the health issues they may be susceptible to (Watson, 2008). Differential diagnoses, especially of genetic syndromes, are associated with specific health issues and knowing these susceptibilities allows families to be prepared and to know what kinds of supports or interventions are potentially going to be required (Watson, 2008).

In the absence of diagnosis, parents may worry about the possibility of reduced life expectancy (Rosenthal, Biesecker and Biesecker, 2001; Watson 2008) and be uncertain about whether their child might eventually die from their condition (Rosenthal, Biesecker and Biesecker, 2001; Lewis, Skirton and Jones, 2010). However, the desire for prognostic information may also be accompanied by considerable ambivalence and while parents may seek to know more about their child's future, they also have concerns about receiving bad news (Rosenthal, Biesecker and Biesecker, 2001). Watson (2008) reports that, although parents sought to know about future expectations, having information about future prognosis could also be quite frightening and not knowing could therefore come as a bit of a relief. Landsman's work with mothers of disabled children (2003, 2009) identifies that, unlike uncertain aetiology that may lead to self-blame, uncertain prognosis may lend itself to anxiety but also hope. This hope is tied to futurity and the notion of progress and not knowing their child's prognosis may leave room for hope that a child may overcome or reduce his or her disabilities.

Rosenthal, Biesecker and Biesecker, (2001) report that some parents said that they wanted to plan for the future, but circumstances had forced them to take it one day at a time. Research work with families with children with chronic, life-threatening illness indicates that parents make use of strategies to deal with sustained uncertainty and the impact of prognostic uncertainty. Cohen (1993) describes how for parents of children with chronic illness, their assumptive worlds are challenged. The management of this near total transformation of the expectations involves strategies including managing awareness of time. Cohen describes parents as tethered to the present and the very proximate future (Cohen, 1993) in the wake of the diagnosis of chronic, life-threatening illness. Parents manage uncertainty by adopting

a 'one day at a time' philosophy and living life in shortened units. This living in the present affects parents' abilities to make future plans, which are associated with a rise in anxiety.

The findings from my own study resonate with the findings from the research work referenced here. Uncertain prognosis was a key area parents felt not having a diagnosis had an impact on. Parents wanted to know about their child's future in terms of potential progress and development and wanted to work in the present towards that future in providing appropriate treatment and therapy. They felt there were no boundaries to what they could expect; they didn't have 'milestones' or 'guideposts' as you might with a named condition. Like the parent in Makela et al.'s study (2009) who referred to seeking the 'enhanced compass' of diagnosis (2009, p2397), parents wanted parameters to what they might expect. Parents wanted to anticipate future threats to their child's health and a small number of parents felt knowledge about life expectancy was difficult without diagnosis. Care in adulthood was also a concern, and parents worried about whether their child would be independent as an adult and who would care for them. As reported in work with families with chronic, life-threatening illness (Cohen, 1993), parents in my own research described using strategies for living with sustained uncertainty like managing time by breaking it down into smaller units and taking one day at a time.

Resonating with other research work with families with disabled children (Rosenthal, Biesecker and Biesecker, 2001; Watson, 2008; Landsman, 2003, 2009), around a third of parents expressed ambivalent feelings towards uncertain prognosis, feeling that the situation of not having a diagnosis was preferable to having a diagnosis with a poor prognosis. The uncertainty of the future caused anxiety for parents, but also allowed for hope about their child's future development.

In this chapter, the significance of diagnosis in looking backwards (to aetiology) and forward (to prognosis) has been suggested. The consequences of living without a diagnosis concerns both of these temporal domains of the past and the future and diagnosis is seen to be important in making sense of the past and in anticipating the future for families. In the next chapter I turn to the domain of the present in considering the day-to-day lived experience of absent diagnosis by looking at the consequences on access to services and support, and the impact on social interaction. I suggest living without a diagnosis is felt to have an impact on

all temporal domains, from making sense of the past, experiencing the present, and anticipating the future.





9.1 Introduction

9.2 Access to services

*Educational Support*

*Applying for Disability Benefits*

*Therapy and other formal services*

9.3 Access to informal support

*Contact with other families*

*Syndrome Without a Name (SWAN) support organisation for families*

9.4 Managing Social Interaction

*Difficulties describing their child's disability to others*

*Strategies for managing social interaction*

9.5 Conclusion

## **9.1 Introduction**

In chapters 7 and 8 I considered how not having a diagnosis was reported by parents as affecting the way they looked both back in time, to what caused their child's disability, and forward in time, to their child's prognosis. In this chapter the temporal domain primarily considered is the present, as I look at how not having a diagnosis was perceived to have an impact on the day-to-day life of the family with reference to three key themes; access to services, access to support, and managing social interaction. Many parents raised these issues spontaneously either when telling their story or responding to my question about how they felt not having a diagnosis had impacted on their lives. Eleven parents spontaneously made reference to an impact on access to services, 12 parents spontaneously made reference to an impact on access to informal support, and 12 parents spontaneously made reference to difficulties describing their child's disability to others in the absence of diagnosis. The remaining parents who talked about these issues did so after being asked specifically whether they thought not having a diagnosis had an impact in these areas. This chapter considers each of these themes. First, I consider access to services; looking at educational support, applying for disability benefits, and access to therapy and other formal services. Then I look at access to informal support in the context of contact with other families. In this section I also introduce the support organisation Syndrome Without a Name (SWAN); further information about SWAN is included in appendix 7. In the final section of this chapter, I consider how parents managed social interaction without the use of a label to aid communication about their child's disability. I explore difficulties parents had describing their child's disability to others, and strategies used by parents to manage these difficulties.

## **9.2 Access to services**

Parents commonly felt that not having a diagnosis had impacted on their child's access to formal services. Twenty-one parents said they felt absent diagnosis had an impact in this way. Three parents felt that not having a diagnosis had not impacted on access to services, and two were not sure. Those parents who felt that not having a diagnosis had impacted in this way described difficulties with: educational support, including support at school and

accessing the right educational placement; applying for benefits, including Disability Living Allowance; and access to therapy services and treatment. Commonly, parents described barriers to accessing services including being unable to obtain services, experiencing delays in accessing services and having to describe their child's disabilities in length due to the absence of a diagnostic label.

### *Educational support*

Ten parents reported that not having a diagnosis had an effect on their child's educational support. Parents were not asked directly if not having a diagnosis had impacted on educational support, although were prompted about impact on services if they didn't raise this themselves. These ten parents felt not having a diagnosis potentially disadvantaged their child with reference to accessing the right school placement, receiving appropriate support at school, or having a formal statement of need that they felt would ensure appropriate support.

Calum's parents felt it was more difficult for the additional needs management team to make a decision on placement and support for their son in the absence of a label.

If it had a label on it, they could look at the label then look in a book and say oh right yeah varying needs for this child could be severe to moderate learning difficulties (Calum's father, 2)

Similarly, Peter's mother linked having a diagnostic label with accessing the right educational placement for her son. She reported that, after being told by the head teacher at the special needs school she wanted her son to attend that it would be easier to get support service for him at school if he had a diagnosis, she took action by going back to her son's consultant and starting 'another round of tests and appointments' in the effort to locate a diagnosis. As part of this process, her son was assessed by a psychologist, who advised her of several diagnoses that she did not feel fitted her son and that were later called into question by other professionals involved in her son's care. Despite disagreeing with the

psychologist's diagnoses, she acknowledged that the psychologist's report had a value in improving her son's access to the school placement she was pursuing for him.

Based on her findings she diagnosed him with autism, ADHD, intellectual disability and [name of diagnosis]. We were devastated and totally disagreed with some of her findings. Nevertheless, at the end of term one he was offered a place in a small special class, so I think the report served a purpose...It was harder to get special education support for Peter not having a diagnosis. He was floundering very badly in the mainstream school system, and we kept being told there were no vacancies in the special classes. It wasn't until the psychologist did give him the label of autism and intellectual disability that they suddenly made a place available (Peter's mother, IF, AUS)

As well as access to the right educational placement, parents also reported access to support in nursery or school was affected by not having a diagnosis. For Ismail's mother, for example, getting a diagnosis was seen as important in getting the right support for him at school and she worried that her son was getting less support than he needed due to not having a 'certain diagnosis'. She felt that as her son only 'classifies as having learning difficulties' (1) he was getting only the minimum level of support. She said; 'children with learning difficulties have the minimum amount of support in school...I need to find out what is wrong with Ismail, so I can push for more support' (1). David's mother also felt that if her son had a diagnosis he'd be getting the support he needed in his education setting. As a consequence of not having a diagnosis, she felt 'his education was a wee bit kind of left to one side' and the support he had was inadequate. She said; 'if he had a diagnosis they would take it more seriously' (2). Without a diagnosis, she felt there was less authority to her requests for support for her son.

As already described, Rupert's parents were in a different situation with reference to desire for diagnosis in that they didn't want their son to be labelled with a diagnosis at the time of the interviews. However, they still had concerns about the impact of not having a diagnosis on their son's support at school. They recognised their son had some difficulties at school that required support, but felt that these difficulties didn't need a specific or unifying label and that it was important to treat the child, not the label. However, they believed it would be easier for the school to access support for Rupert if he had a specific diagnosis. This led to concerns that 'the plug would be pulled' (1) on plans for classroom assistance if they did not pursue diagnosis or accept diagnoses suggested by the educational professionals involved.

These examples may raise the question of whether access to support is improved if parents are prepared to compromise in accepting a diagnosis they may believe to be inappropriate. For Peter and Rupert, at least, it seemed that a diagnosis was a potential gateway to improved support at school or a better educational placement. The significance of diagnosis in the education setting, as potentially a necessary tool to secure support, may see parents accepting a diagnosis that they do not see as accurate. In this way, Peter's mother 'totally disagreed' with the diagnoses offered by her son's educational psychologist, yet realized that they activated appropriate support for her son; 'they suddenly made a place available, where non-diagnosis couldn't' (IF, AUS).

Some parents linked support in their child's educational placement with having a formal record of their child's needs (now referred to as a 'Co-ordinated Support Plan' in the UK, but at the time of the interview study was still called a 'Record of Need'), which was perceived to be more difficult without a diagnosis. Eva's mother, for example, felt very strongly that Eva was not offered a 'statement describing her educational needs' because she didn't have a diagnosis. Because of this she felt that her daughter had not been properly acknowledged by the local authority as a child with disabilities and was therefore 'in danger of missing out on provision to which she may be entitled to' (IF, ENG). She described a struggle to obtain services and support. Cameron's mother also worried that diagnosis had an impact on 'opening a record of needs' (1). She felt that with a diagnosis, a child would be given a record of needs that would record the additional help that they needed and provide a legal obligation to give that support to the child. She felt there was less legitimacy to the demand for support without a diagnosis.

Ten parents reported that not having a diagnosis had an impact on their child being able to access the right educational placement or the right support at school. This finding suggests the importance of category, in that a diagnostic category recognized by the school or local authority may make it easier for support to be put in place. Without the shorthand of a category, there may be more of a reliance on description. It is suggested that a diagnostic category, particularly a well-known label, may mean more in this context than a description of a child's needs. While for all diagnoses there is a spectrum of how affected a child may be, without a specific diagnosis this spectrum may be perceived as being much wider. The use

of very general labels like 'developmental delay' or 'learning disability' or 'muscle disorder' may mean little when trying to access specific educational support, while more specific diagnoses may be seen to open doors to support.

### *Applying for disability benefits*

Five parents reported that not having a diagnosis had an impact on applying for disability benefits. Several parents spoke at length about this. Parents were not asked directly about applying for disability benefits so parents raised these issues spontaneously. For these 5 parents, it was the process of applying for Disability Living Allowance (DLA) that was reported to be affected by not having a diagnosis, with one parent further identifying applying for prescription glasses to be a further difficulty without diagnosis.

Emily's parents described how they had to 'fight their corner' to access disability benefits for their daughter in the absence of diagnosis. Without a diagnosis, the administrative task of applying for benefits was reported as being more difficult.

What it feels like is that it's harder to cross those bits of red tape because you don't have a definitive diagnosis of what the problem is (Emily's mother, 2)

Emily's father described trying to access disability benefits as being 'most frustrating'. Without a diagnosis, he felt the credibility of their application for DLA was affected.

Not having a diagnosis I mean that was a blooming nightmare, it was as if you were making the whole thing up (Emily's father, 2)

He felt they had to fight for support all the time due to lack of diagnosis and that they had been refused higher level DLA, until they took it to appeal, because they didn't have a diagnosis. He said not having a diagnosis 'makes it more difficult' and experienced frustration that there was nothing 'to put down' on the forms.

I mean that's one of the major problems we have I mean obviously when you've got a child that's got special needs eh you apply for benefits and things like that and you know it's like well what do you put down? Nothing wrong? (Emily's father, 1)

Emily's mother and father suggested that the need for a label in itself was important, regardless of whether that label was well understood.

The optician actually said that she had like cerebral visual impairment so [Emily's father] thought oh right that's good to put on the DLA form, you need to have a big something that looks wordy or whatever and folks look at it and think oh right okay we've got that...you know in essence for folk like DLA and some places does it really matter if you could tell them that you had some weird syndrome that had just been developed then they may well just accept that...I mean obviously [Emily's father] had said enough to put them off asking any more (Emily's mother, 2)

So a diagnostic label, even if obscure, is seen as 'enough' for those administering disability benefits; a label would give credibility to the application. Emily's mother felt that without a diagnosis 'there's nothing to write down' and that the 'whole story' needed repeated again and again. Not being able to write down a concise diagnosis was felt to be a problem in applying for benefits like DLA, and also an on-going problem for obtaining a prescription for glasses for her daughter. Again, Emily's mother felt that using specific diagnostic language would assist the process of applying for disability benefits and that labels opened doors to this kind of support.

It's still a problem in terms of her glasses prescription ... normally you'd be able to write down oh you know the child has autism or down's syndrome and folk would look at it and go right okay that's fine but with Emily you have to write the whole story...it's still another way that I have to fight the corner (...) Doors are opened when you say your child is autistic which will never be opened if you say my child's got special needs and development delay cause that's as much as we know (Emily's mother, 2)

Calum's parents and Caroline's mother described similar difficulties with applying for DLA. Calum's father described how their claim for DLA was rejected because his son 'didn't fall into any of the categories'. He was convinced that DLA was initially refused because his son didn't have a specific diagnosis. He felt that it had been difficult for those administering the process to 'tick the box' and award DLA.



We got a letter back from them saying he doesn't fall into any of the categories go away sort of thing...not having a diagnosis had an impact absolutely without a doubt...The impression we got from the Department of Work and Pensions was...they're sitting down with a sheet with some boxes and they need to tick the boxes and because Calum doesn't fall into any of the prescriptive categories they went oh fine [makes the action of throwing something away] (Calum's father, 1)

Caroline's mother recalled how she used a label of 'congenital ataxia' in applications for disability benefits, although she didn't see this label as a diagnosis.

These examples bring out the potential importance of diagnosis as an administrative category in terms of accessing resources, and the felt consequence of not having a label to put 'in the box' when applying for disability benefits. In the case of those parents who felt their application for DLA was refused as they didn't have a diagnosis, absent diagnosis is seen to have serious consequences in their ability to access resources. As with support in education, it could be that parents who make use of labels to *act* as diagnoses (like 'cerebral visual impairment' or 'congenital ataxia'), even if they doubt the accuracy or specificity of such labels, may more easily access resources or support.

#### *Therapy and other formal services*

Nine parents also reported that not having a diagnosis had impacted on access to therapeutic support and other formal services for their child or family. They believed their child was not accessing the most effective therapies and treatment because they didn't have a diagnosis.

Parents made general reference to difficulties accessing support in this way, as well as specific references to therapies and interventions. The kind of services and formal support that was affected by not having a diagnosis included access to: occupational therapy and physiotherapy services (Calum's mother); speech therapy (Eva's mother, Cameron's mother, Emily's mother); social work services (Eva's mother, Cameron's mother, Ismail's mother, Caroline's mother, Emily's father); treatment (Jessica's mother); and equipment (Sarah's mother)

She has undergone therapies that have been highly distressing for her that had we known what it is that she has we probably wouldn't have tried (...) If we had a diagnosis we would have a better handle on what we should do for her, what are the most important things for us to target? What type of therapy is most likely to be successful? (Jessica's mother, IF, AUS)

If I could tell them [social worker] a diagnosis that might help, because you know a lot of things like autism people are aware of it...so they have some understanding of it (Ismail's mother, 1)

In this section I have introduced data that suggests that a significant number of parents perceived not having a diagnosis had an impact on access to services with reference to education educational support, applying for disability benefits, and accessing therapy and other formal services. Difficulties obtaining services for families with disabled children are well documented. Parents often have a constant 'battle' to obtain services suited to their situation and needs (Beresford, 1995). Having to fight for services can increase stress for parents of disabled children (Campbell-Hall, Coulter, and Joyce, 2009), and unmet need for support from services is identified as an important factor associated with coping and related to parental well-being (Sloper and Turner, 1993). With reference to formal support services, Clavering (2007) reports inertia in improvements in service provision in the UK for families with young disabled children and suggests that parents continue to experience barriers to care and support. Many parents may struggle as much with coming to terms with fragmented service provision as with their child's difficulties (Clavering, 2007). Clavering and McLaughlin (2007) highlight that service procedures are opaque and are not responsive to human need and McLaughlin et al. (2008) suggest difficulties with accessing support services may still be commonly experienced by families with disabled children. Although the ethos for provision of services to disabled children and their families in the UK is one that recognizes need over diagnosis, the majority of parents in my own study (21 of 26) perceived that not having a diagnosis had impacted on their access to formal support and services.

### 9.3 Access to Informal Support

Most parents (20) felt that not having a diagnosis had an impact on their access to informal support. A small number of parents (2) felt absent diagnosis had not impacted on access to informal support, and 4 parents were ambivalent or were not sure whether there had been an impact in this area. Those parents reporting that not having a diagnosis had an impact on their access to support felt that making contact with other parents of children with the same or similar difficulties was more difficult without a diagnosis. Parents found it difficult to make direct contact with other families or to make contact with other families by joining a relevant support group.

#### *Contact with other families*

Ten parents specifically reported that they wanted to make contact with other parents in a similar situation to their own but found it difficult to do so without diagnosis.

[With a diagnosis] we would be able to get in touch with other people with the same diagnosis for information and support. I would love to be part of a formal support group with families with similar children (Peter's mother, IF, AUS)

Being able to learn from other parents saying you know my child has this and we've found this works to deal with whatever, I've never really had that (Ismail's mother, 1)

It would be better if we had a diagnosis...we could find other kids other families that had gone through the same thing (Moy's mother, TI, USA)

It was difficult for parents to be in touch with families directly, or to join a relevant support groups. Absent diagnosis was seen to impede contact with other families. Again, some parents compared their own situation in this respect to those of parents of children with common diagnoses.

With no set diagnosis you couldn't turn up to any group of people to ask for help or support. If your child's been diagnosed with Down syndrome you can approach a particular group of people or with autism ... you know the support groups and all

these kind of things but you're just left to get on with it if you don't have a diagnosis that's the bottom line (Emily's mother, 1)

I mean you get groups for autism and you get groups for Down syndrome but when you don't have any diagnosis at all, I mean you don't fit into any wee sort of pocket anywhere you know (Emily's father, 2)

Calum's father perceived a clear support mechanism was in place for parents of children with a named diagnosis, but missing for parents of children without. This support mechanism was described as already established, ready to be accessed by families with named diagnoses.

If you had a diagnosis I think there's a mechanism there already...you get a diagnosis for that there's loads of help groups, Contact a Family and things like that are all there so you've got this support mechanism in place and you know exactly where you're going and your future path is laid out for you and you can see the path relatively clearly and you can move on (Calum's father, 1)

He recognised the therapeutic value in being able to access a support group, without which he believed it was possible to feel alone. He felt not having a diagnosis had meant they had missed out on that support.

It's always nice to go to these things and realise that there's other people in this boat as well, so you're not alone, so just for that (Calum's father, 1)

Abdul's mother spoke of community when talking about absent diagnosis and the impact on parent-to-parent support; 'it's just being part of a bigger group really...you know you're automatically part of that larger community [when your child has Down syndrome or autism], the community of people who have children like Abdul is much smaller' (2). Like Emily's father (you don't 'fit into any wee pocket anywhere'), for Abdul's mother absence of diagnosis meant not being able to fit easily in to a support community. She described in the past how she was 'desperate to form a connection somewhere...desperate to talk to anybody who would understand' (1). She recalled going through a 'low' period in the early years of her son's life and how she was 'desperate for some contact'.

Jessica's mother spoke at length about feeling that a sense of community was missing in the absence of diagnosis. She craved a sense of belonging (to 'know where we fit' IF), a need to

not feel alone in her parenting journey; 'to know that there are other people out there like us and that there are support groups for people like us and there are people who know what it feels like to *be* us we are not alone (...) a sense of belonging...of having other people journey with us' IF. As well as the importance of being able to join a support group for families with the same diagnosis, making contact with other parents online is also suggested as valuable.

I could jump online on the internet and see there'd be forums on the computer...there'd be chat rooms and there'd be resources in the library and support groups....I need you know I'd just like to talk to other people who are experiencing similar things (Jessica's mother, TI, AUS)

She described how she was still actively seeking this connection; 'I'm looking around trying to gravitate towards people who are like us' (TI) and how these efforts were made difficult by not having a diagnosis.

Two parents described their experience of accessing a more general support group for parents of children with a range of disabilities. Cameron's mother had attended a general support group for parents of children with special needs.

I feel when I go to the parents' support group there are parents of kids with the same diagnosis...maybe Autism or Asperger's, you know they can sort of speak to each other and maybe give each other practical tips, but there's absolutely not really anything like that [for me] (Cameron's mother, 1)

Peter's mother explained that she used to attend a general support group but over time everyone else got a diagnosis and started to attend their 'speciality' support group. She felt that without a diagnosis they were unable to access valuable support and there was no-one they could relate to who had the same problems.

For parents of children who do not have a specific diagnosis for their difficulties, being unable to make contact with other families may lead to feelings of isolation.

By missing that connection it makes me feel more isolated than included (Moy's father, IF, USA)

Many of the parents (20) felt that not having a diagnosis had impacted on their access to support. These parents felt that it was difficult to make contact with other families with children with the same or similar difficulties either directly or via a support group. There was the sense of something missing for these parents; a sense of community, belonging somewhere, being able to connect with others by virtue of shared experiences. Absent diagnosis had implications for parents' emotional well-being in that support was often desired, but not available to them.

The challenges faced by parents of children with disabilities and complex health problems in terms of unmet need for psychological and emotional support have been widely reported (Beresford, 1995; Sloper and Turner, 1993; Lindblad, Holritz-Rasmussen, Sandman, 2007). Parents of children with a specific diagnosis may find it easier to join or form associations than parents of children with a vague or completely uncertain diagnoses (Lenhard et al., 2005) and having a diagnosis can improve access to contact and with support from other families (Howlin and Asgharian, 1999). As parents of disabled children are reported to experience higher stress levels than parents of non-disabled children (Sloper and Turner, 1993), access to adequate support is important. Lack of parent support and high levels of parental distress may affect not only the parent but also the child's well-being (Sloper, 1998). Olsen et al. (1999) suggest that one of the main factors affecting hardiness of families of disabled children is access to social support. Stress may be reduced when parents are able to share and compare their experiences with other parents who are in a similar situation and parents of disabled children may be uniquely qualified to help each other, with contact with other parents potentially having a positive transforming effect and helping to eradicate feelings of isolation (Kerr and McIntosh, 2000). For many parents in my own study, this vital parent-to-parent support was absent as, without a diagnosis, it was difficult for parents to make contact with other families with children with similar difficulties.

*Syndrome Without a Name (SWAN) support organisation for families*

At the time of carrying out the interview study, there was one UK based internet and phone-line support organisation set up to offer support to families 'living without a diagnosis':

SWAN (Syndrome Without a Name). In appendix 7 I include information about this support organization, both as it was set up at the time of the interview study and as it exists now, and report data from the interview study relating to parents' knowledge and/or experience of SWAN. Very few parents in the interview study had heard of SWAN or made use of its services (only 3 of the UK based parents), although one of these parents described making contact with other families via SWAN as particularly useful and beneficial.

An interview was carried out with the original founder of the SWAN support group in the UK (also discussed in Appendix 7). The founder of SWAN had unique contact at that time with parents living without a diagnosis and a good understanding of their perceptions of absent diagnosis. She met with parents, had email and written communication with them, and personally manned the SWAN helpline that parents called for support and advice. She also had direct experience of a family member being severely affected by a disability that had no conclusive diagnosis. As she was not the parent/guardian of a child without a diagnosis, I did not include the data from her interview in the data analysis elsewhere in the thesis, but I felt it was important to acknowledge the issues raised in our interview. In this interview, she identified the key areas that parents felt not having a diagnosis had an impact on as being: accessing to formal and informal support, including access to benefits, educational support and equipment, and making contact with other families; on knowing what caused their child's disability and being able to assess the risk of recurrence; and on having information about their child's prognosis. She said that parents often experienced feelings of isolation. She also talked about the phrase 'swan child' and how she had acted in different ways to try to get this phrase accepted as a medical term. Our interview helped to clarify the kind of support provided by SWAN at the time of the interview study, and also was a form of triangulation of the themes that emerged from the interview study with parents in that many of the areas identified by parents in my own interview study as being affected by not having a diagnosis, were commonly raised by parents in contact with SWAN.

In this section, I have suggested that many parents had unmet need for support, and that contact with other families and joining a relevant support group were difficult without a diagnosis. Although at the time of the interviews a support organization was in place for

families living without a diagnosis, many parents hadn't heard of the organization which was fairly small, relied on the work of a small number of volunteers, and was based in the founder's own home in Staffordshire which had limited resources and was far from where the parents in the interview study lived. With little support from and contact with other families, parents may feel isolated. In the following section I consider an issue that may add to this sense of isolation in looking at difficulties parents experienced with social interaction, in that many parents struggled with managing definitions of their child in social situations.

#### **9.4 Managing Social Interaction**

Parents commonly reported that not having a diagnosis had an impact on describing their child's disability to other people. Twenty-one parents reported experiencing difficulties managing definitions of their child in social interaction. Parents described particular difficulties with managing social interaction with people they didn't know; some difficulties describing their child's disability to family and friends were also reported.

##### *Difficulties describing their child's disability to others*

Parents reported not knowing what to say to other people to explain to their child's disability ('if we knew what was wrong it would make it easier to explain to people' Calum's mother, 1). They felt they had to give a lengthy rather than a 'snappy' answer (Calum's mother); that they had to give a very detailed description of their child's difficulties rather than a concise diagnosis; ('It makes it very difficult to explain clearly and concisely what is wrong with Peter', Peter's mother, IF, AUS); and that other people didn't understand what was meant by an 'undiagnosed syndrome' or didn't understand the notion of not having a diagnosis.



What's wrong with her? And you're well right okay how long have you got! (...) I mean it is just difficult to to say to people you know if you're in a shop or something the kind of situations where Emily's not known (...) it's amazing people are very surprised they're very surprised people that haven't had this experience find it astonishing that nobody's you know told you what it is and I suppose really when I was on the other side of it I would probably have been the same (Emily's mother, 2)

Here, Emily's mother highlights the low cultural resonance of non-diagnosis, people haven't heard of it, and admits that she would have been as unaware herself had she not experienced absent diagnosis.

Parents described going into lots of detail when responding to questions about their child's disability, which was potentially time consuming and unsettling. Jessica's mother, for example, described how herself and her husband struggled with 'doing the whole undiagnosed thing' (IF), sometimes listing four or five characteristics of their daughter's condition: epilepsy, ataxia, hypotonia, hypertonia and intellectual disability, in the absence of a unifying diagnosis. She said:

It's not simple, it's messy...diagnosis affects this massively people ask what is wrong with Jessica and we just look at each other and wonder where to start (Jessica's mother, IF, AUS)

Abdul's mother similarly said she did not have any concise explanation for her son's difficulties to use in social situations; 'I don't think I have an answer that I use...I just blurb [sighs]' (1), and Anna's mother reported that she often gave different explanations and wasn't comfortable with any one of them; 'This is a problem for me all the time. I'm always saying something different, then I just feel uncomfortable' (IF, USA). Without a concise diagnosis, going into lots of detail may have seemed the only option when asked by members of the public.

We don't have a name for it and it's hard to tell people and people say what's wrong with her, how can you tell them? You go into all the details (Claire's mother, 1)

One parent noted that the problem was experienced with medical practitioners and members of the public alike.

When people not familiar with Moy come into contact with her and ask what her syndrome is and we have no answer a perplexing or glazed look comes across their face...It doesn't matter if that person is a doctor, pharmacist or stranger in the grocery line (Moy's father, IF, USA)

Several parents worried that other people didn't believe what they were saying when they described their child's difficulties in the absence of a clear diagnosis ('I always feel people don't believe me because I can't say 'he has this'' Peter's mother, IF, AUS). Four parents commented on how others possibly didn't believe their explanation and two parents suspected that others thought their child was being badly behaved ('they see him as a misbehaving child' Cameron's mother, 2). Several parents felt that others may judge them for not having a diagnosis, thinking they were uncaring, strange or lazy.

It's difficult not having a diagnosis because you have to go through all the ins and outs of what's wrong with them...they seem to think you're a weird mother (Andrew and Lee's mother, 1)

Because she looks quite normal people don't understand why she screams and bites herself and we certainly have had comments about being bad parents or her being a naughty child trying to get attention and what have you (Jessica's mother, TI, AUS)

Absent diagnosis may have emotional impact, and receiving comments about their child's behaviour or their parenting in social situations may further augment feelings of isolation. In these cases, parents experience felt stigma because they don't have a diagnostic label with which to explain their child's behaviour or development difficulties.

Four parents worried that other members of their family would have difficulty describing their child's difficulties to others. They anticipated the negative impact for their parents or parents-in-law and/or their other children.

I feel our daughter suffers because she as well isn't able to tell her friends 'she has this' so it's very difficult for her friends to understand what she goes through (Emily's father, 2)

More than half of parents referred to strategies for managing social interaction in the absence of a diagnostic label, including avoiding social situations, using general or umbrella labels to explain their child's difficulties, and using a diagnosis they believed to be inaccurate.

Two parents described avoiding social situations or interactions that would necessitate having to give an explanation of their child's difficulties.

I don't like going out with David it's the stress factor with me going out I don't tend to say to people now I used to give them an explanation I tend to just run away as quickly as possible (David's mother, 2)

And I know [Moy's mother] tries to avoid those situations because she is tired of repeating the same story. We have become less social. And less for me means non-existent (...) When you tell someone your child has Down Syndrome they get a knowing look on their face, when you tell someone we don't know what her problem is we get a lot of dumb looks. Sometimes to the point where you think that person believes you aren't doing anything for your child. I just try not to describe anything to anyone I am becoming more antisocial all the time (Moy's father, IF, USA)

David's mother and Moy's father describe avoiding giving explanations, either by leaving social interactions or rarely going out. Moy's father perceives he is judged by others for not acting to find diagnosis for his child and highlights that conditions with high cultural resonance, like Down syndrome, are widely 'known' and understood. His words again highlight the possibility of the felt stigma that may be associated with not having a diagnosis for your child's significant difficulties.

Around half of parents (13) made use of general diagnostic terms or umbrella labels to explain their child's difficulties to others.

You just have this big difficulty trying to explain what's wrong...development delay, I suppose we just latched on to that because it was the only thing we had, because what do you say other than that? (Emily's mother, 2)

In making use of these general labels or umbrella diagnoses, parents were aware they were not defining their child's disability accurately but felt it was easier to manage social interaction in this way.

We do kind of use the partial diagnosis overgrowth syndrome you know we've kind of gotten around it that way (...) We've cottoned on to that we just tell people he's got an over-growth syndrome (...) alright it's a general diagnosis but we use that to explain to everybody what's wrong with him (Calum's father, 2)

The most common general label used by parents in social encounters was 'development delay', with about a third of all parents using this label to help manage social encounters. Only one of the parents used the phrase 'undiagnosed syndrome' (Jessica's mother) to explain her child's disability to others and another used the phrase 'swan child' (Sarah's mother).

Three parents made use of a label that was not their child's diagnosis in social interactions but was a diagnosis with high cultural resonance, therefore giving their child's disability potentially more legitimacy and helping others understand that their child was disabled. Caroline's mother, for example, used the diagnosis of 'cerebral palsy' in social situations so that others could make sense of her daughter's disability. She recalled how they had initially been told their daughter may have cerebral palsy. They told family and friends that this was their daughter's probable diagnosis. When they realised this wasn't the case, they choose not to update some 'wider family and friends'. They relied on the diagnosis of cerebral palsy, even when it was ruled out, as a way of avoiding difficult explanations. Cerebral palsy was seen by Caroline's mother as 'a label I could use' (1) with family and with people she met in social situations.

Similarly, David's mother described how she used a label with strong cultural resonance (autism) to relieve the difficulties of managing social interaction. She said:

I have at times said he's autistic because I think it's hard to say he's got behaviour problems and he can't understand what you're saying, so if you say to people yeah he's autistic, a lie, they kind of grasp it because a lot of people know what autism is (David's mother, 1)

Although she was aware that telling others he was autistic was a 'lie', it was an effective diagnosis for managing social interaction in the absence of diagnosis. In her second interview, she described avoiding social interaction with people who did not know her son, as she no longer wanted to fabricate an explanation.

One other strategy used by just one parent was telling others their child was younger, so that people's expectations of their child's development were managed. Moy's mother described how she once told people she had met for the first time that Moy was younger. She said 'I'm always tempted to just tell them a lie and say she's only 1' (TI, USA), however she had only used this explanation once. By managing other people's expectations of her child's development on this occasion, she didn't have to give difficult explanations or explain that they didn't have a diagnosis.

Around half of parents made use of general diagnostic terms or umbrella labels to explain their child's difficulties to others and a small number of parents made use of a diagnosis they believed to be inaccurate when explaining their child's difficulties to others. Other research with families has indicated that a diagnosis is important in dealing with the public and helping others to understand a child's behaviour (Gillman, Heyman & Swain, 2000). A label can be an explanatory device used by families of disabled children used to 'explain' their child (Makela et al., 2009). A diagnosis is an important interactional resource to be used in day-to-day situations (Gill & Maynard, 1995), a tangible entity utilised to fence other people's questions (Brookes-Howell, 2006). Brookes-Howell, looking at the experience of families with children with genetic disorders, reported that for families of children without a clinical diagnosis, an 'everyday' diagnosis (for example, 'autism') may be used in their interactions with others. This diagnosis may empower the parent in their social interaction with others, as an 'interactional resource' to be used in a day-to-day setting (Howell-Brookes, citing Teas Gill and Maynard, 1995). Diagnosis is held to be a social resource, useful in talk with others. Different settings for diagnosis are identified, the clinical and the everyday, as well as different modes of diagnosis, *informing* in the clinic and *talking* in the everyday (Brookes-Howell, 2006).

There has been little focus on the experiences of parents of disabled children in public spaces (Ryan, 2005a, 2005b, 2008) and barriers to inclusion for families may be attitudes and

responses of others to disabled children in public spaces (Ryan, 2008). Ryan (2008) looks at how mothers of children with learning disabilities make use of labels in public spaces to account for their child's behaviour. In one off interactions (with no past or future), mothers did not necessarily want to engage in lengthy explanations of their child's difficulties. Many mothers relied on the term 'autism' to explain their child's behaviour to others and so aid social interaction in public spaces. Autism was used as a shorthand to manage public interactions, even if this label was not diagnostically accurate. Mothers made use of the simplest and most cost effective (in terms of time and understanding) method they had to manage social interaction. Ryan (2008) concludes that mothers do tend to provide accounts to explain their child's behaviour in public places, saturated as public places are with rules governing appropriate behaviour. Some mothers were flexible in their use of medical terms and appropriated the most useful term to assist social interaction.

Similarly in my own research with families, many parents were flexible in their use of medical terms and appropriated umbrella or general labels, and in a small number of cases common diagnoses, to manage social interaction. A small number of parents also described avoiding social interaction because of the difficulties of explaining their child's disability. In chapter 6, I suggested that non-diagnosis has little cultural resonance. This impacts on social interaction as people parents meet for the first time may not understand why their child has significant developmental, learning or behavioural difficulties yet no explanatory label for these difficulties. Parents tried to manage these difficulties using a number of strategies. Undiagnosed disability, with little cultural resonance, may be experienced as socially stigmatising. Drawing on Goffman's (1963) theory of stigma, it seems that in avoiding social situations or using an umbrella or incorrect diagnosis to explain their child's disability, parents act in 'covering' (Goffman, 1963) the potentially stigmatising status of non-diagnosis. Lack of diagnosis is concealed, and social encounters managed, by making use of labels that are less mysterious and less likely to cause confusion.

## 9.5 Conclusion

Many parents perceived that not having a diagnosis had impacted on access to services (21 parents) and informal support (18 parents), and in managing social interaction (21 parents). These findings are largely consistent with findings from other research work with families, although the latter finding was not reported in earlier work with families.

Parents of undiagnosed disabled children may perceive lack of diagnosis to contribute to difficulties accessing support and services (Rosenthal, Biesecker and Biesecker, 2001; Genetic Alliance, 2010; Lewis, Skirton and Jones, 2010; Makela et al., 2009; Watson, 2008; Graungard and Skov, 2007). Parents may view getting a diagnosis as a way of improving access to services and treatment, leading to earlier intervention and treatment for their child (Makela et al., 2009), gaining knowledge about appropriate interventions and access to funding or specific services (Watson, 2008), accessing appropriate intervention (Howlin and Asgharian, 1999) and as a way of accessing help for their child or being able to do more to help their child (Graungard and Skov, 2007; Williams, 2007). Parents may seek diagnosis in the belief that it will be followed by treatment and intervention, along with social support (Gilman, Heyman & Swain, 2000). Parents may have concerns that their child is not accessing the appropriate treatment in the absence of a clear diagnosis and may feel that a diagnosis would lead to a better overall strategy for treatment of physical and developmental problems (Rosenthal, Biesecker and Biesecker, 2001). Lewis, Skirton and Jones (2010) reported that one of the reasons parents wanted a diagnosis was to get help with schooling. In another study with families with undiagnosed children, parents perceived the value of diagnosis lay in being able to access help with care and treatment (Genetic Alliance, 2010). Parents felt that if they had a diagnosis it would enable them to make more accurate decisions regarding their child's care and treatment and difficulties associated with not having a diagnosis were reported as impact on accessing; financial help, educational support, and receiving help from social services and health professionals (Genetic Alliance, 2010).

Parents without a diagnosis may feel isolation (Rosenthal, Biesecker and Biesecker, 2001; Genetic Alliance, 2010; Lewis, Skirton and Jones, 2010) and view their situation as unique. Even those parents with an awareness of the high incidence of unidentified syndromes may

feel isolated (Rosenthal, Biesecker and Biesecker, 2001). Rosenthal, Biesecker and Biesecker, (2001) identified a strong desire among the parents to affiliate with a larger community of parents. Some parents of children without a diagnosis may participate in non-specific support groups for children with special needs, although may wish to find a support group better matched to their child's difficulties (Rosenthal, Biesecker and Biesecker, 2001). Families without a diagnosis may only be able to access 'generic' groups rather than groups specific to their child's condition (Genetic Alliance, 2010). When parents who were members of the support organisation SWAN UK were interviewed (Genetic Alliance, 2010) reasons commonly given for joining SWAN UK included finding support and reading about and meeting others in similar situations. Parents talked about reading other families' stories, having someone to talk to, gaining information about testing, and having a home that gave them a sense of belonging (Genetic Alliance, 2010).

Parents of children without a diagnosis may have unmet need for support from other families (Rosenthal, Biesecker and Biesecker, 2001; Genetic Alliance, 2010; Lewis, Skirton and Jones, 2010; Makela et al., 2009; Howlin and Asgharian, 1999; Lenhard et al., 2005). Parents may seek emotional support by wanting to contact other parents, with mothers potentially being more likely to express a desire for social support than fathers (Rosenthal, Biesecker and Biesecker, 2001). Reasons for seeking others may include a need for emotional companionship, understanding and sharing information (Makela et al., 2009). In several studies parents of children without a diagnosis compared their own situation to that of parents of children with well-known conditions like Down syndrome when talking about social support and contact with other families (Rosenthal, Biesecker and Biesecker, 2001)

Parents in my own study reported that not having a diagnosis had impacted on access to informal parent-to-parent support and that they had difficulties connecting with other families because they did not have a diagnosis. Parents of children who do not have a diagnosis may feel they do not belong to any specific 'community of suffering' (Helman, 1978), although they may wish to feel part of a larger community of parents. This may have a negative impact on those parents who feel this support may benefit them. Parents expressed a desire to affiliate with a larger community of parents, but did not have access to such a community and didn't feel they 'fitted' anywhere in that respect. This was described by some parents as leading to feelings of isolation. This sense of isolation, and the need for



emotional companionship, information sharing, social support, contact with other parents and families, and the ability to form associations and relevant connections, is in keeping with findings from research with other families living without a diagnosis (Rosenthal, Biesecker and Biesecker, 2001; Lewis, Skirton and Jones, 2010; Genetic Alliance, 2010; Makela et al., 2009; Gilman et al., 2000; Howlin and Asgharian, 1999; Lenhard et al., 2005).

Parents in my own research commonly reported that they had difficulties describing their child's disability to others; this finding was not reported in earlier work with families with undiagnosed children in which difficulties describing their child's disability to others was not directly identified as a significant theme (Rosenthal, Biesecker and Biesecker, 2001). However, in this work it was reported that parents saw the value in labels as a way to help others understand the condition (Rosenthal, Biesecker and Biesecker, 2001). In a later study with families living without a diagnosis (Lewis, Skirton and Jones, 2010), carried out after my interview study, a small number of parents (3 of 14) suggested the importance of diagnosis in social interaction. The authors report that diagnosis was seen as a social tool by a number of parents, who had difficulty describing their child's disability to others (Lewis, Skirton and Jones, 2010). These parents felt it would be useful to have a term that could help to summarise what was wrong with their child when asked by friends or other parents and a small number of parents used general terminology ('special needs' and 'development delay') instead of referring to undiagnosed disability (Lewis, Skirton and Jones, 2010, p808). Lewis, Skirton and Jones (2010) describe this as using a 'social diagnosis' to manage social interactions in the absence of diagnosis (Lewis, Skirton and Jones, 2010, p810). In a similar vein, Madeo et al. (2012) highlight that 'knowing what to tell others' (p1882) can be a difficulty in the absence of diagnosis, and Makela et al. (2009) reported that parents felt a diagnosis would establish proof of a credible problem and provide their child's siblings with words to explain their child's intellectual disability to others. Using a diagnosis in social interaction can validate a child's condition and confirm the 'reality' of it, and give clarity without the need for further explanation (Avdi et al., 2000).

The extent of the difficulties parents encountered describing their child's disability to others was unexpected based on prior literature. The majority of parents reported difficulties, particularly with people they met for the first time. Without a diagnosis, parents did not have a shorthand concept to explain the nature of a child's difficulties to others. Parents

recognised that certain diagnostic terminology (including umbrella or general labels or common diagnoses like 'autism') had high cultural resonance and were therefore useful. This finding highlights the utility of a diagnosis for a parent; how without a diagnostic label parents may experience barriers in social fields as they negotiate social settings without the capital afforded by a recognisable label.

This chapter has considered aspects of the day-to-day lives of parents of disabled children without a diagnosis and the impact of absent diagnosis on access to services, support and on social interaction. Access to informal support and managing social interaction, in particular, may be uniquely difficult for parents of undiagnosed children compared to children with more well known diagnoses. Access to services and formal support is dependent on funding and availability, and all families with disabled children may struggle to access the formal support they need. Not having a diagnosis may make that process more difficult or time consuming. Contacting other families with children with the same condition is, however, ruled out without diagnosis, as is having an accurate label to use in the management of social interaction. Very rare conditions, or conditions without a medical label, have low cultural resonance and consequently make it difficult for parents to be understood when telling others about their child's disability and to be in contact with others with similar difficulties.



## Chapter 10 Discussion and Conclusion

### *Discussion*

This thesis tells multiple stories revealing a play of diagnosis journeys: that of the parent participants living without a diagnosis; that of the researcher's own journey; and that of the sociological significance of diagnosis. A different origami could have been made with these stories, for there is variety in the sequence of the folding. My own story, and an identified gap in the literature, was the catalyst for researching this topic. That there was little empirical work looking at the experience of families living without a diagnosis, and that absent diagnosis in this context was under-theorised, was the impetus for me choosing to explore an experience that seemed to have high prevalence among families with disabled children and significant consequences, yet little attention. My own biographical disruption, in a sense, was the beginning of this thesis journey, and perhaps the thesis was my acting possibility, or even my coping strategy. In a sense, I ran after a truth after all.

As already described, the mutual sharing element of the interviews was less significant than I had anticipated. I did not really share my story with any of the participants, because I was there to hear their story, although the mutual stock of knowledge we shared may have helped to form the bedrock of an understanding between us. And while qualitative research is fundamentally subjective, I felt I gave the participants as much opportunity as possible to tell me their own stories. Although parents were guided to talk about areas of interest largely generated by previous research and theory, these questions followed from an uninterrupted telling of their stories and an open-ended question about whether not having a diagnosis had an impact on their lives. Many of the issues raised were done so spontaneously, as I have described in each data chapter.

The structure of the presentation of the data analysis in this thesis is similar in structure in some respects to the recent work in the sociology of diagnosis in that I consider the different components of diagnosis; diagnosis as process (the parents' quest for diagnosis in chapter 4), followed by the consequences of absent diagnosis (in chapters 6 through to 9). Additional analysis is carried out and presented in chapter 5, exploring parent narratives. When data

analysis began in 2007, prior to the publication of recent sociology of diagnosis work, I structured analysis by conceptually dividing the data into two areas: the search for diagnosis and the impact of non-diagnosis. I also considered classification and the way medical categories were created and structured. In later drafts, following the publication of key sociology of diagnosis work in 2009 and 2011, I endeavoured to create a three-tiered structure adding in the dimension of diagnosis as category. However, I could not make 'diagnosis as category' fit to inform the structure of the data analysis, unable to separate it as a distinct part forming one of the domains of the thesis. Instead, it permeated all parts of the thesis. Families had an enduring engagement with category in their prolonged journey through the process of diagnosis. Indeed, it was a category of diagnosis they invested much time and energy in looking for; it was the missing object of their quest. Their journey in search of a medical label was a journey in search of diagnosis as a meaningful category. The chapter I had headed 'Diagnosis as category' remained almost empty of data analysis, including instead a plethora of literature. This was not because diagnosis as category was not important to the parents' experiences, but because it permeated their experience. I suggest that while it is worthwhile to consider the different components of diagnosis as distinct, it is important not to reify the distinctions between them so completely that their mutual relations are overlooked. These components bleed into one another, and their relationship is circuitous, as Jutel acknowledges (Jutel, 2011a). In trying to structure the thesis into the domains of diagnosis as process, diagnosis as category and (non-)diagnosis as consequence, I learned that this structure may not be suitable for all topics relating to diagnosis.

In this thesis I have suggested the importance of narrative and offered reasons why it is worthwhile to consider the whole story of individual parents as a complement to discussing parsed extracts of their interviews in the context of themes across narratives. Thematic analysis is still held to be crucial; narrative analysis can complement our understanding of the experience of living without a diagnosis by considering the whole story of individual participants and, in a sense, letting their stories *breathe* (Frank, 2010). Parents told teleological stories, of a journey seeking a diagnosis. For almost all parents, some transformation was described as they made sense of living without a diagnosis. In a sense there was no resolution to their quest, although commonly a degree of closure in their diagnosis journeys

was suggested, along with the suggestion that parents remained open to possibilities for diagnosis in the future.

The theme of story and journey permeate the thesis. These motifs are seen in data collection, as parents are asked to share their stories and tell their diagnosis journeys; in my approach to the literature and in finding the right methodology as these reviews are taken on as an intrinsically interpretive process, hermeneutic and iterative; in data analysis, as I consider and adapt Frank's (1995) narrative typologies; in considering the *process* of diagnosis that parents participated in; and in the core metaphor of a journey halted in its movement identified in parent narratives. A story of my own journey is also woven through this thesis.

Parents told stories that resonated with the quest motif in literary narrative. Absent diagnosis was suggested to represent biographical disruption, the villain who disturbed the peace. There is a dual aspect to this experience of fracture for parents, as both their child being disabled and further being unable to account for that disability with diagnosis is suggested to represent biographical disruption and a fracture of parents' expectations. After establishing their child had significant difficulties that required a medical explanation, the parents' journey commonly involved extensive searching for a definitive diagnosis. In keeping with both the literary quest motif and the quest typology in studies of illness, parents are searching for something that is missing, a lack. They begin their search with the belief it can be found. Like the seeking of an emerald, a homeland, a grail, a child turned into a swan; their journey's goal is to find what is missing and bring it home.

That the parents in my own research commonly sought diagnosis with such intensity is a finding that contributes to existing knowledge from the small number of studies examining the parental experience of living without a diagnosis. In this literature a parental 'quest' for diagnosis is identified (Rosenthal, Biesecker and Biesecker, 2001; Maher, 2013; Graungard and Skov, 2007; Watson, 2008, 2009), a 'diagnostic odyssey' (Reiff et al., 2012). McLaughlin et al. (2008), in their work with parents with disabled children, suggest that a common narrative, especially for mothers, is the battle to obtain a clear diagnosis after their child is born or when their differences become apparent. In their quest, parents may seek diagnosis 'in the privileged grand narrative of medicine' (McLaughlin and Goodley, 2008, p323).

Parents in my own research told stories of a drive to find an answer to their diagnosis question. The drive to find the definitive answer may be reflective of western culture's biomedical drive and symptomatic of our solution demanding culture (Davis, 1960). The parental drive to solve the diagnostic puzzle may represent a reaction to mystery that is culturally typical. As Gordon (1995) suggests, the majority of us living in the modern industrial world share an anxiety about the unknown and the uncertain to the degree that our reaction to mystery is to drive towards the imposing of order and control. Our faithfulness to the scientific paradigm, that the world is ultimately explainable, may be played out in our reaction to unknown diagnosis, particularly when what is at stake is as important as our child's wellbeing. McLaughlin and Goodley (2008) raise concerns that those who speak on behalf of marginalised groups may make assumptions that create overly simplistic conceptions about the individual or the community, for example with reference to the assumption that parents of disabled children need a high level of certainty (see also Goodley, 2007a, 2007b). McLaughlin and Goodley (2008) describe parents of disabled children as both nomad and settler, in that they 'carve out changing ways of living, which engage with shifting moments of certainty and uncertainty' (2008, p318). Over time, parents in their study negotiate their experience by seeking to ensure that their child does not become 'contained, categorised, subjectified' (2008, p327) within a diagnosis, embracing new forms of uncertainty in their lives and their visions of the future. There is a sense in their work of parents resisting diagnosis that was not as strongly present in my own research. Although parents in my own research became more tolerant of living without a diagnosis over time, they still desired diagnosis and there was little expression of negative views towards obtaining a definitive diagnosis. Ambivalence towards certainty of diagnosis was expressed in discussion about prognosis, however commonly parents described an enduring (if reducing) desire for diagnostic certainty. In short, *knowing* was perceived as better than *not knowing*. The caveat for this was when knowing meant discovering that your child had a diagnosis with a very poor prognosis. If parents increasingly accepted uncertainty, they did so not by entirely giving up on the seeking of diagnosis, but by accommodating uncertainty alongside the unresolved quest for diagnosis, a quest that was tempered but not surrendered. I suggest that accepting the uncertainties inherent in diagnosis can co-exist with continuing to be open to the possibility of obtaining a diagnosis in the future and that, in this way, modernist and postmodern agendas co-exist (see McLaughlin and Goodley, 2008; p331).

Parents were active in the search for diagnosis. From a place of diagnostic limbo, they were agentive in the search to find a definitive diagnosis for their child. They used multiple methods to seek diagnosis, with the internet being the primary tool for search activity. The internet was used to make sense of signs and symptoms, research tentative diagnoses, find new possible avenues for diagnosis, and to seek others to help in the quest for diagnosis and for support. The process of diagnosis involved parents' accessing, assessing and reappropriating medical knowledge (Nettleton, 2004), as they utilised e-scaped medicine (Nettleton, 2004) in their search. As the one who knew their child best and with a high level of motivation, yet a relative lack of experience or training in medicine, they occupied a unique position relating to the process of diagnosis; driving the process with a high level of motivation for diagnosis, carrying out their own work of diagnosis, and negotiating tentative diagnoses with their child's consultant. Despite the difficulties associated with searching for diagnostic information, the internet was a valuable tool for parents enabling them to take part in the diagnosis process as contributors to the work of diagnosis. Their research activity in the process of seeking diagnosis relied on expert knowledge and experience to fully make sense, and so their diagnosis work was carried out in conjunction with the work of the clinician. Thus, while a feature of their agency was that they carried out interpretative work themselves, it was suggested that medical knowledge consists of 'something in addition to the facts' (Millar and Wynne, 1988, p112) and that the 'craft expertise' (Millar and Wynne, 1988, p112) of their child's consultant was needed to allow them to fully interpret their findings. There was an emotional element to parents' engagement with diagnosis, and the risk of emotional harm as parents considered multiple possible diagnoses. Different diagnoses were considered, and parents experienced false summits as they researched the appropriateness of different conditions before ruling them out and starting their search again. Considerable emotional and intellectual resources were needed for the search and tension was suggested between the diagnostic search as constructive and hopeful yet, after a time, destructive and hopeless.

In their active participation in the work of genetic diagnosis, both in and outside of the clinic, parents practiced a kind of *lay dysmorphology*. They carried out practical observation (of their own child and others) and reasoning (in interpreting whether diagnostic information was relevant or not) and looked for patterns in symptoms and signs (Shaw et



al., 2003). Parents endeavoured to 'fit' their child into a particular syndrome or diagnostic label through a process of alignment and exclusion characteristic of diagnosis work in the dysmorphology clinic (Latimer et al., 2006). They carried out some of the nosological work of diagnosis. In trying to solve the diagnosis puzzle, they sought to dismiss diagnoses that were a poor fit for their child's signs and symptoms, and kept on diagnoses that their child fitted well with. Their search for diagnosis involved a process of 'pattern recognition' (Hunter, 2002). In carrying out this lay dysmorphology, parents enacted a remarkable level of participation in the process of genetic diagnosis.

Parents made sense of the process of diagnosis with syncretic understandings (Santos, 2006), fusing lay with scientific understandings. It is suggested that by participating in the process of diagnosis parents may learn to 'talk the talk of the clinic' (Latimer 2007a, p113), passing through a process of proto-professionalisation (Shaw, 2002, Hilton and Slotnick, 2005) as they actively participate in diagnosis. One feature of their agency was the way they negotiated diagnosis by taking the results of their search activity back to the clinic, sometimes challenging the doctor's own diagnosis work. At our particular intersection of the information and genomic age (Driessneck, 2014) parents, in the face of absent diagnosis, are likely to engage in the diagnosis process by accessing information online. The expert clinician is needed to interpret the results of the parents' findings; thus in a sense remains the arbitrator of knowledge, an essential link for the parent practicing lay dysmorphology.

Parents diagnosis work was indicative of the practice of dysmorphology not only in their observation of the signs, symptoms and features of their own child but also in the seeking of other children who resembled their own. As identified in other work looking at the process of paediatric genetic diagnosis (McLaughlin, 2014; McLaughlin and Clavering, 2011) parents looked for other children who resembled their own, through face-to-face contact (for example Jessica's mother looking at other disabled children she worked with) and by looking at photographs of other children with genetic conditions online (for example Moy's father 'looking into these kids eyes' or Eva's mother looking for others like Eva, asking 'Could that be Eva? Has anyone else got an Eva out there?'). Parents also assessed for similarity and difference in written accounts of other children's conditions. At times, the *gestalt* of being able to see the face of a syndrome (Latimer, 2013) was suggested, for example by Sarah's mother when she explained that she liked to *see* other children rather than just

comparing their symptoms to her daughter's, because even when a child shared the same clinical characteristics on paper, the look of the children could be different: 'sometimes our children have very similar symptoms in words but are rather different in appearance'. The visualisation practices of genetic diagnosis occur not only in but also beyond the clinic (McLaughlin, 2014), which parents in my own research enacted by analysing images of other children for both recognition and connection.

In around half of the narratives of parents who consulted with geneticist, a theme of genetics as an advancing field was identified. In parent narratives genetics was played as *becoming*, at an intersection in terms of being on the brink of significant advance that are not yet actualised. It is only since advances in technologies for genetic testing have come about in very recent years, with next generation sequencing technology and an ability to detect new genetic conditions through whole exome sequencing (Turner et al., 2009; Choi et al., 2009; Worthy et al., 2011), that it is possible to put into context the advances that parents may have been alluding to. For all parents, this perception of genetics as an advancing field (what Latimer calls 'a new frontier of knowledge', 2007b, p14) was tied to conversations that had taken place in the genetics clinic, although parents may have supplemented their understanding about genetics with information found outside of the clinic, like Abdul's father did. This finding suggests that the geneticist's own knowledge about diagnosis or perception of genetics as an advancing field acted as a reference through which the parents drew meaning in the diagnostic encounter (McLaughlin, 2005) and is a further example of the way parents made use of syncretic knowledge to understand their experiences, fusing the knowledge or suggestions they encountered in the clinic with their own everyday understandings.

In light of anticipated future scientific advances, although diagnosis was not possible at the time it was perceived as potentially being possible in the future. In this way, many parents perceived diagnosis as deferred. A rhetoric of improving technology and a commitment to the progress of science was identified in parent accounts. Latimer (2007a, 2013) suggests that the clinic is seen as a space of deferral, and that parents may be shifted between definition and deferral in their engagement in the clinic (Latimer, 2007a, 2013). On the one hand they are engaged in a prolonged process of seeking definition for their child, and on the other hand they are aware that limits in current technology may make this definition unavailable

in the present. The notion of deferral of diagnosis may reassure parents, keep hope for a diagnosis alive, and allow them to temper their eager quest and, in doing so, redirect the time and energy they were spending searching for a diagnosis.

The findings of my own research also suggested that with diagnosis deferred, many families were kept on at the clinic (Latimer, 2007a, 2013). The benefits of attending the clinic for parents were not limited to medical benefits. The caring work of the clinic was both medical and pastoral. I suggest that while the clinic may provide an opportunity to speak positively of the child, reassure parents they are doing everything right, and alleviate feelings of blame for parents, in particular mothers who may be more susceptible to self-blame (Featherstone, Gregory and Atkinson, 2006), it may also be a more abstract space of hope and connection. Knowing there is someone there, someone to turn to with questions or concerns, were reasons suggested for continued attendance at the clinic. The clinic may act as an anchor (as Emily's mother suggested) for parents, without which they may feel 'set adrift'; perhaps deferral, ambiguity, and uncertainty may be tolerated if moored with the notion that in the future diagnosis may be achieved. Hope is permitted by both the playing of diagnosis as deferred and being kept on at the clinic. Aase (1990), and later Shaw et al., (2003) and McLaughlin and Clavering (2012), refer to the 'watchful waiting' that occurs across time in the genetics clinic. The parents in my study referred to this same process as 'wait and see'. What is interesting is that both phrases 'watchful waiting' and 'wait and see' incorporate both time passing and the visual element to the process of genetic diagnosis.

A small number of parents, when talking about genetic testing, considered genetic diagnosis through the lens of heredity and expected genetic testing to offer conclusive results ruling out or in a genetic aetiology. In this way, parents' prior beliefs did not always synchronise well with the scientific explanations they encountered (Driessnack, 2014). A larger number of parents, around a third, were aware of uncertainties in genetic testing and the complexities of genetic causation and had understandings of genetic attribution that went beyond explanations of heredity. As already suggested, the development of new sequencing techniques has the potential to dramatically alter the diagnostic landscape for families (See Dimond, 2014b). Currently in the UK, families of children with unexplained developmental disability who fulfil study criteria can access whole exome sequencing in a clinical research capacity. This was not available at the time of the interview study, although advancing

technologies were clearly suggested by parents. In a sense, the data from my research can be used to consider how parents may make sense of knowledge-in-the-making. They expressed syncretic understandings of concepts they encountered in the genetics clinic, understandings that were accommodated in their continuing to live without a diagnosis.

Parents commonly experienced a decrease in focus on diagnosis and in search activity over time. In describing parent narratives I adapt Frank's (1995) typologies and identify three common typologies in parent narratives: *restitution and the new normal*, *times of chaos*, and *unresolved quest*, with *unresolved quest* being the dominant typology. In being unable to find a diagnosis, parents experienced a kind of liminality, in that they were in a diagnostic no-man's land; living in lag time land. Unlike Turner's (1987) liminality, however, parents were involved by a process of participation rather than subjection (Latimer, 2007a, p113; Latimer, 2013). They participated actively in the work of diagnosis and moved to varying degrees beyond the state of being betwixt and between by accepting absent diagnosis, readjusting previous assumptions about the world, and to an extent learning to incorporate the uncertainties that came with absent diagnosis into their daily lives. They needed to 'go on', as Jessica's mother said, for themselves, for their child, for their family, for their community. Their search often took them away from their child, family, or work; it was time-consuming, tiring, and potentially upsetting. For a variety of reasons a general trend of parents' search activity and thinking about diagnosis reducing over time was identified. This finding resonates with other research work with families with undiagnosed children (Rosenthal, Biesecker and Biesecker, 2001).

For parents in my own research, the passing of time was identified as a factor in the felt need for diagnosis reducing. The unresolved quest, once they 'got off the train' (Emily's mother) of so intensively seeking diagnosis, became a kind of status quo. The quest did not end, but there was a hiatus in search activity and of having diagnosis in the foreground of their lives and thoughts. This lull could be disrupted with change, in their child or in their knowledge, and the search was again invigorated. In this way, the diagnostic quest ebbed and flowed; with a general trend of reducing over time in intensity. I described self-transformation with most parents coming to accept living without a diagnosis; thus gaining new insights on their journey. This transformation varied between parents, and was not complete in that the door to diagnosis was left open. So a tension remained at play between their acceptance of living

without a diagnosis and the possibility for the search for diagnosis to be reanimated at any time. Not all parents experienced self-transformation in accepting absent diagnosis. Even for parents who did express self-transformation, their attitudes were complex. It was not that they wanted a diagnosis and then, as time went on, they no longer did; there was ambivalence and contradiction amidst a general trend of the felt need for a diagnosis decreasing over time. Furthermore, a metaphor of stasis in relation to movement was suggested powerfully in many narratives. This may have suggested the difficulty of the journey for parents, as movement through the process of diagnosis was halted, obscured, made more difficult. This core metaphor may even represent a contradiction in the narratives of many parents as self-transformation was described yet, at the same time, stasis implied. I suggested letting that contradiction, if it is one, be. In doing so, I hope to remain faithful to the complexity of the families' life worlds.

In considering the consequences of living without a diagnosis, it was suggested that absent diagnosis has consequences across all temporal domains; the past with reference to how parents thought about cause; the present with reference to access to services and support, and in managing social interaction; and the future, with reference to assessing the risks of recurrence and knowing what to expect in the future. The key findings of my study with reference to the consequences of absent diagnosis contribute to knowledge about the lived experience of parents of disabled children who do not have a diagnosis. Many of the findings reflect the findings from earlier work with families living without a diagnosis (Rosenthal, Biesecker and Biesecker, 2001; Lewis, Skirton and Jones, 2010). The research also makes new contributions to empirical work and new theoretical connections.

All parents speculated about cause, with most mothers and one father expressing self-blame, often accompanied by associated feelings of guilt. A discourse of risk characterised these expressions of self-blame and thinking about cause; with most mothers and one father considering what they may have done or not done to cause their child's disability. Mothers commonly referred to their conduct in pregnancy, with reference to idealised notions of good mothering and correct behaviour in pregnancy. This finding resonates with other work in this area, like Landsman's research with mothers (2009), extending this work into the domain of absent diagnosis. My findings suggest that, in the absence of diagnosis, 'deviance' (Goffman 1959) associated with responsibility for the onset of their child's disability may

never be abrogated, particularly for mothers who culturally are held to carry more of the responsibility for pregnancy outcome. Without a clear diagnosis, there are no boundaries to individualised blame for adverse outcomes. Actions or omissions of the parent cannot be ruled out and the parent cannot prove or disprove their culpability. My findings regarding parents' attitudes to aetiology are in keeping with the findings from other research work with families that identify cause as an important aspect of not having a diagnosis for parents (Rosenthal, Biesecker and Biesecker, 2001; Lewis, Skirton and Jones, 2010) and supports suggestions that parents may be more likely to blame themselves for their child's condition when there is no clear answer regarding the cause of their child's difficulties (Nixon and Singer, 1993). Not knowing aetiology also had an impact on future reproductive choices: with many parents choosing not to have another child because of the unknown risk of recurrence. A small number of parents did go on to have more children, basing their decisions more on 'gut feeling' than scientific evidence of risk of recurrence.

All parents referred to the future or their child's prognosis as uncertain, with many parents specifically linking this uncertainty of prognosis with not having a diagnosis. It was suggested that we all live with uncertainty, but for parents of disabled children without aetiological or prognostic certainty, the world may seem even more unpredictable. Again, this finding is consistent with other work with families in this area (Rosenthal, Biesecker and Biesecker, 2001; Lewis, Skirton and Jones, 2010; Avdi et al., 2000; Makela et al., 2009; Madeo et al., 2012). As previously identified by Rosenthal, Biesecker and Biesecker (2001), parents in my own research at times expressed ambivalence about the desire for prognostic information, concerned about receiving a diagnosis with a poor prognosis. Parents also developed strategies, like breaking down time into smaller units, for dealing with uncertain prognosis. In this way, like families with chronic life-threatening illness described in other research, they may have remained tethered to the present and the very proximate future (Cohen, 1995), despite desiring information about the past and the future.

In considering the consequences of absent diagnosis on the day-to-day lived experience of families, I suggested that not having a diagnosis was perceived by most parents to have an impact on access to services, access to informal support, and in managing social interaction. Diagnosis as an administrative category was seen to be useful in accessing educational support and in applying for disability related benefits, and some parents felt their request

for support had less validity without a recognised label for their child. Parents sought contact with other families and described an unmet need for such contact, often expressing a sense of isolation and a need to belong or 'fit' somewhere. Connection with others was felt to be missing in the absence of diagnosis. Again, these findings regarding support and services are consistent with other research work with families looking at the significance of diagnosis (Rosenthal, Biesecker and Biesecker, 2001; Genetic Alliance, 2010; Lewis, Skirton and Jones, 2010; Makela et al., 2009; Graungard and Skov, 2007; Gilman et al., 2000; Howlin and Asgharian, 1999; Lenhard et al., 2005). It is suggested that parents of children who do not have a diagnosis may feel they do not belong to any specific community of suffering (Helman, 1978) which may lead to feelings of isolation. This isolation may be compounded by difficulties they have in social interactions. The extent of the difficulties parents encountered describing their child's disability to others was unexpected based on prior literature. The majority of parents reported difficulties, particularly with people they met for the first time. Without a diagnosis, parents did not have a shorthand concept to explain the nature of a child's difficulties to others. Parents recognised that certain diagnostic terminology (including common diagnoses) had high cultural resonance and were therefore useful. This finding highlights the utility of a diagnosis for a parent; how without a diagnostic label parents may experience barriers in social fields as they negotiate social settings without the capital afforded by a recognisable label. I suggest that in having unmet need for informal support, and encountering difficulties managing definitions of their child in social interaction, the peripheral membership that may be felt by families with disabled children may be more acutely experienced in the absence of diagnosis.

Further to these perceived consequences of living without a diagnosis, I also described how parents had difficulties making sense of absent diagnosis. The idea of unattainable diagnosis being difficult to understand was a feature of many parent narratives, and absence of diagnosis was expressed as bewildering and resulting in a kind of existential or epistemological anxiety. Families had to redefine their subjectivities when faced with absent diagnosis, an experience that I suggested may have fractured their assumptive worlds (Janoff-Bulman, 1992). Parents' assumptions and expectations about the world, in particular about medical science, were potentially challenged by absent diagnosis, with the experience of absent diagnosis potentially exposing the limitations of medical knowledge and practice (Kelly, 2007, p94). High expectations of medical science and a perception that medical

science is achieving ever more complex feats may give rise to difficulties making sense of being unable to achieve diagnosis, with absent diagnosis incongruent to the high profile achievements of medical science ('they can clone animals...and yet why can't they tell us?'). Furthermore, as non-diagnosis has low cultural resonance and most parents were unfamiliar with undiagnosed childhood disability prior to their experience, they may have experienced a fracture of their expectations and epistemic shock (Kelly, 2009) that diagnosis was not possible. This may have been compounded by finding out in a moment of disclosure that a diagnosis may not be possible. Conditions with high cultural resonance, Down syndrome and Autism Spectrum Disorders, were often framed by parents as representing a contrast with their own situation of absent diagnosis. I suggest non-diagnosis as a natural breaching experiment, exposing our assumptions about medicine and beyond. Although other research with families has highlighted parents' 'need to know' (Rosenthal, Biesecker and Biesecker, 2001; Lewis, Skirton and Jones, 2010; Graungaard and Skov, 2007; Madeo et al., 2012; Lenhard et al., 2005; Makela et al., 2009; Genetic Alliance, 2010; Brookes-Howell, 2006), the conceptual difficulties potentially associated with the experience of living without a diagnosis have not been a key consideration in this work. My research suggests that absent diagnosis may be experienced as bewildering, as previously held assumptions are fractured and that the loss parents experience with the fracturing of these assumptions may be ambiguous.

In the literature review chapter, I suggested that much of the work in the newly emergent sociology of diagnosis is concerned with diagnosis as category, in that it looks at classification in medical diagnosis, the social framing of diagnosis, contested and medically unexplained conditions, and drivers of diagnosis (Jutel, 2011a). Much of this work concerns medicalisation. I suggest that for parents of undiagnosed children, their own experience of being unable to obtain a diagnosis for significant disability may also be experienced as bewildering and incongruent to the cultural drive of increasing medicalization in all realms of life. The enduring engagement families with undiagnosed children have with categories of diagnosis is inconsistent with current directions in the scholarly work on medicalisation. That is, the experience of non-diagnosis is at odds with the contemporary dialogue of ever increasing medicalization of everyday life (Szasz, 2007) which may represent a kind of cognitive dissonance for those experiencing absent diagnosis. If it is the normal that is itself shrinking (Latimer, 2013) then how do increasing tendencies to diagnose impact on an



experience in which significant life-limiting disability is present yet a diagnostic label absent?

My work on diagnosis is also different to that of the current work in the sociology of diagnosis in that it studies a complete *absence* of diagnosis for conditions that are agreed upon as medical. Absent diagnosis is seen to hold a mirror to diagnosis, telling us more about what diagnosis *means*. In the same way that breaching experiments (Garfinkel, 1963) can expose the rules of behaviour norms, so too can absent diagnosis expose the meanings of diagnosis and our assumptions and expectations about diagnosis, medical science or even our understandings about the world. Here, I suggest that absent diagnosis exposes the fault lines of, and opens an analytical window to, the multiplex meanings of diagnosis. It is suggested that *absent diagnosis for uncontested medical conditions* may be seen as a new area of diagnostic tension; new in that it is currently under-theorised and that there are gaps in empirical work looking at the lived experience of negotiating absent diagnosis in this context. As signs and symptoms for those patients who do not have a diagnosis for significant impairment are extremely varied, looking at absent diagnosis in these circumstances tells us much about what diagnosis means across a range of human disorders of development. This thesis therefore contributes to knowledge about diagnosis, and extends current work in the sociology of diagnosis, by considering this area of diagnostic tension.

In considering categories of diagnosis and medical classification systems in the literature review and beyond, I suggested that residual categories may not be meaningful to parents. Patients occupying a space of 'residual other' in medical classifications systems may have been overlooked from a research perspective, despite prevalence being high (Fairburn and Bohn, 2005). Not Otherwise Specified categories may be considered a kind of Cinderella state, a garbage label (Bowker and Star, 2000) that does not make sense for families. The ICD-10 (WHO, 2004) contains an abundance of residual categories, covering it seems all eventualities of morbidity and mortality. In this way, it is possible to assign a medical code to all illness or disorders of development. In the 10th revision of the ICD (WHO, 2004), for example, a code of Q99.9 would refer to 'chromosome abnormality, unspecified'; code Q89.9 to 'congenital anomaly NOS' and 'deformity NOS'; code R99 to 'death NOS' and code R69 to 'unknown and unspecified causes of morbidity' or 'illness NOS'. Armstrong (2011) highlights the difficulties of applying a classification system of disease to 'the rawness of

undifferentiated human illness and distress' (2011, p801). For conditions agreed upon as medical but for which there is no precise label, the 'not otherwise specified' categories are the only option for medical categorisation. Medical classification systems such as the ICD or the DSM are information infrastructures not commonly accessed by parents, and being able to assign a child a code of a residual 'not otherwise specified' category may not be meaningful to families. I suggest that undiagnosed childhood disability may expose the fault lines and fractures in the current medical classification systems, despite those systems already accounting for the inherent uncertainty of diagnosis with the inclusion of residual sections.

In the literature review and beyond I have also made reference to medically unexplained symptoms (MUS). MUS as a label has been identified as a 'polemical knot'; a thingamyjig (Greco, 2012, p2364). I suggest that the label 'Syndrome without a name' (SWAN, or Swan child) carries a similar placeholder function. The acronym SWAN is akin to MUS in that it is not a diagnosis, does not represent homogeneity of patient group, and may have limited meaning. Like MUS, its truth is 'an absence of truth' (Nettleton, 2006, p1176). Corbin and Strauss (1985) suggest that when diagnosis is not secure, there is diagnostic limbo. I suggest that the label SWAN, like the label MUS, does not free a patient from this diagnostic limbo and does not perform any of the expected functions of diagnosis. The container category of SWAN loses much of its meaning in its elasticity. 'Swan children' may differ radically in signs and symptoms, and some of the benefits a precise diagnosis may bring (like making contact with other families with children with similar difficulties or describing their child's disability to others) are not possible with the pseudo-diagnosis of SWAN. Whilst SWAN has little cultural resonance and lacks homogeneity of patient group, it will have little meaning as a diagnosis. It is a gentle sounding way of saying that a child does not have a diagnosis, but if relied on as a diagnostic label by parents there is the risk that it reifies the idea of a homogenous group of children described by a diagnostic label. For this reason, I assign 'Syndrome without a name' a question mark in the title of the thesis. I initially used this term in the title because it had a degree of lay presence and, because my work looked at the lived experience of parents, I wanted to reflect lay definitions of absent diagnosis. However, as I learned, very few parents in my research were aware of the term or the support organisation of the same name (at the time of the interview study). Parents sought a precise and

meaningful diagnosis; 'syndrome without a name' did not function as, or have the meaning of, a diagnosis.

Considering that all morbidity and mortality can be assigned a medical code and label within current medical classification systems, and that there exists terminology to describe even those syndromes which have no name, gives rise to a central question for this thesis of what is being referred to when we talk about absent diagnosis? What were the parents seeking? What did they perceive to be lacking? Diagnosis in this context is a more complex notion than simply whether the clinician has provided what is thought to be a diagnosis or not, but instead concerns what parents consider to be a diagnosis. The data from this research and the small number of other studies with families living without a diagnosis has suggested common areas of diagnostic meaning and significance. A medical code is not enough. A medical category is not enough, when that category is a residual or umbrella term that is not perceived to explain all of a child's difficulties. A unifying label that will account for the potentially 'extraordinary disruption' (Latimer, 2013) that many children experience in their development is sought. And further than this: this diagnostic label must have *meaning*. It is an accepted medical category, it is an explanation of cause and effect, it is a social resource. In a sense, parents in this research were seeking the benefits associated with common rather than very rare diagnoses. Perhaps this is why so many parents made reference to diagnoses with high cultural resonance, like Down syndrome and Autism Spectrum Disorders, playing these more common childhood diagnoses as antidote to their own uncertain diagnostic situation. The attractiveness of obtaining a diagnosis for parents was to an extent associated with childhood conditions commonly identified before genomic medicine, and also with childhood disorders like Autism Spectrum Disorders that are not necessarily associated with genetic aetiology but are well known. Relatively common diagnoses like Down Syndrome and ASD have high cultural resonance; they offer the ability to look backwards to cause (in the case of Down Syndrome at least) and forwards to prognosis, to manage social interaction, to access support and services, and they also make sense in that their occurrence is familiar. This is not necessarily so with rare or newly identified genetic diagnoses. New and rare genetic diagnoses may not have these meanings, or at least they may not have these meanings *yet*. Are the benefits or certainties parents perceived diagnosis may bring, given that new genetic diagnostic tools may alter the meaning and content of diagnosis, therefore illusory? Given the prolonged nature of the

testing process that had already been carried out, it is likely that their child would receive a future diagnosis of a very rare disorder rather than of a well known disorder. Would the diagnosis of a very rare genetic disorder bring with it the benefits and certainties that parents felt they lacked?

It appears that parents were seeking a definitive diagnosis for their child for a number of reasons. They sought diagnosis so that they may better understand what caused their child's disability and, in some cases, so that they may potentially alleviate self-blame. Yet even for parents who receive a diagnosis, speculation or self-blame may not be entirely alleviated. They sought diagnosis so that they may make better informed future reproductive choices. Yet even with named genetic disorders, risk of recurrence may be an estimate and there may be no guarantees. They desired knowledge regarding appropriate treatment and interventions, and sought a label in order to access services; yet a diagnostic label does not necessarily bring with it easy access to social work, education and health services and support and parents of children with named diagnoses may yet play warrior and advocate for their child. Contact with other families relies on a community of others with shared experience, which may be difficult in cases of very rare conditions. And in managing social interactions, the utility of a label may rely on known shared definitions. Uncertainties about a child with a genetic condition's health, development, cognitive abilities and life span often persist even after diagnosis (Stewart and Michel, 2000; Rosenthal, Biesecker and Biesecker, 2001; Lenhard et al., 2005). Parents of children with rare genetic disorders would be expected to have a high level of perceived uncertainty due to the limited prognostic information because they may have only been reported in a few individuals (Lipinski et al., 2006). The meanings and benefits parents perceive they may lack in the absence of diagnosis may only be associated with well known childhood disabilities. It may be illusionary, therefore, that a future diagnosis for their child may mean knowing the past and predicting the future, having a shorthand term for social interaction, and accessing support and social resources. A rare genetic diagnosis may be a shorthand summary very few outside the genetic community may understand (McLaughlin and Goodley, 2008). The journey seeking meaningful diagnosis for parents of children who have already travelled far down the diagnostic funnel may be a journey of illusion, like the Sanskrit *Maya*, that-is-not, but which appears to be real. And as the seeking of the name may have high costs in terms of time,

energy and intellectual and emotional investment, there may be an element of destruction produced by this illusion of certainty.

Some parents voiced an understanding that the diagnosis of a rare genetic disorder would not grant them the information or benefits that diagnosis of a more common disorder would. Perhaps one runs after it all the same. The space of motility that parents are identified as occupying, between deferral and definition, is potentially a space of ambiguities and contradictions. They journey on an active quest, and yet their narratives are replete with metaphors of stasis. Did the stasis in their journeys lead to an increasing acceptance of uncertainty over time? Were they, like the parents in the work of McLaughlin and Goodley (2008), seeking certainty about various aspects of their child's lives while at the same time embracing new forms of uncertainty? Parents may be thus both nomad and settler (McLaughlin and Goodley, 2008); able over time to experience self-transformation in coming to live without certainty of diagnosis, yet holding on to hope for diagnosis in the future and as such revealing narratives potentially forever in search of meaning (Hyden, 1997). By taking each day or each developmental stage of their child's life at a time parents' narratives were evocative of Bauman's continuous present (Bauman, 1996), yet parents did not let go of their desire for a certain, definitive, biomedical diagnosis. Parents' active and eager quest and strong desire for diagnosis upholds the normative structure, reflecting a world dominated by the hegemony of normalcy (Davis, 1997) and where taxonomy is a dominant paradigm for understanding this world. Yet at the same time, they transformed to varying degrees to accept uncertainty of diagnosis.

The suggestion that the benefits and certainties of diagnosis are illusionary can also be challenged. It may be that for very rare diagnoses, there are still some benefits shared with the more common diagnoses with high cultural resonance. It may be a question of degree, with more common diagnoses offering clearer parameters and rare diagnoses offering lesser boundaries around what to expect and less information. A diagnosis, while not being able to offer certainty about what they future may hold, may offer parameters about what to expect, a spectrum of possibilities. Should only a handful of other people around the world have been identified with that diagnosis, there may still be more information available than without a diagnosis, albeit limited. Parents may make contact with those other families for the purposes of prognostic information and also to fulfil a need for solidarity, of finding

others that may potentially know what it is like to *be* them, as Jessica's mother phrased it. One of the families in the study was in touch with me soon after the interview study when they received a genetic diagnosis. Moy was diagnosed with a very rare genetic disorder that, at the time, only a few people around the world had been identified with. Moy's father had eagerly immediately been in contact with the other families, keen to connect and share knowledge and experience.

Parents also sought diagnosis in order *to know*. There may be cognitive benefits of a diagnosis beyond its utility in terms of information, support and services. In other work with families living without a diagnosis the importance of knowing is emphasised (Watson, 2008; Makela et al., 2009) and in Watson's study (2008) many families could not articulate the need to know and their stories incorporated cognitive factors related to the families' search for meaning. The importance of knowing and having a name may go beyond the need for explanations of cause and effect and the social resources a diagnosis may offer. Obtaining a diagnosis may give parents peace of mind. Frank (1995) highlights that in restitution narratives the nature of suffering changes from a mystery to a puzzle. A mystery can only be faced up to; a puzzle can be solved. In accepting diagnostic uncertainty and telling narratives more closely akin to quest than restitution narratives, parents take on board that the puzzle cannot currently be solved and, to varying degrees, measure up to and take on board the mystery of diagnosis. Here, I ask whether it may be possible to hold onto both the mystery and the puzzle of diagnosis. Parents may in time come to accept the mystery of absent diagnosis, and live to varying degrees with diagnostic uncertainty, yet in the background they may retain the puzzle and their wish to solve it. Finally obtaining a diagnosis would be finishing the puzzle, giving them cognitive peace.

A final consideration regarding this question of what we refer to when we talk about living without a diagnosis is whether the notion of living without a diagnosis would be different now, in 2014 and beyond, to 2005 and 2006 when the interview study took place. As new genetic technologies advance, particularly as public awareness of these technologies rises, then the landscape for considering absent diagnosis will change. Perhaps now and in the future it will be even more commonplace for families with children with unexplained developmental disorders to perceive that they live with diagnosis deferred, rather than absent. The cultural resonance of undiagnosed conditions may rise as we become more

aware of the ability of new genetic technologies to identify and name disease-causing genetic mutations. Perhaps the mystery that many families have transformed to accommodating in their lives by learning to live with uncertain diagnosis, albeit potentially ambivalently, will transform wholly back to puzzle. If whole exome or genome sequencing becomes in time a cost and time efficient way of discovering diagnoses, and becomes reliable and less complex to interpret then perhaps, to paraphrase Abdul's father's, one day it will be possible to put the information into a machine and out pops an answer. What that diagnostic answer will offer however, in terms of meaning, is uncertain.

A recent online article concerning a newly identified rare genetic diagnosis poses the question 'When is genomic sequencing worth the cost?' It is suggested that obtaining a diagnosis, albeit the diagnosis of a very rare genetic disorder, has the benefit of ending the diagnostic odyssey for the family. Even in the cases when there may be no cure, treatment, little notion of prognosis, and limited understanding of cause, there may still be a value to diagnosis in that it marks the end of a family's diagnosis odyssey. The family are able to move on. Very few parents with a child developing differently refuse to go through a diagnostic journey (McLaughlin and Goodley, 2008). Obtaining a diagnosis would be the end to that journey of searching; it would be reaching Ithaca. On their diagnostic odyssey the parent has perseverance and realises an acting possibility for their child by being agentive in the search for diagnosis. The experience of self-transformation on this odyssey as they accommodate diagnostic uncertainty over time brings a kind of end to their journey, sees them reaching the haven of the Ithacan harbour. Obtaining a diagnosis may be to lay anchor and step ashore.

### *Limitations and Difficulties*

In this section I consider the limitations and difficulties of the study. An important consideration for the research is that parents voluntarily responded to adverts about the research. It must be considered that, in self-identifying as 'living without a diagnosis', the responders may have been different to non-responders. This is an issue for all qualitative research and impacts on the generalizability of the study. I acknowledge that parents who volunteered to take part in the research may have, by virtue of identifying themselves as living without a diagnosis, differed in the way they thought about diagnosis from those who didn't. I do not suggest that results of data analysis in my research are generalizable to all parents of disabled children. However, absent diagnosis was clearly a significant issue for parents involved in the research and it may be so for other families. As findings from data analysis in my own research were similar to findings in other research with families (Rosenthal, Biesecker and Biesecker, 2001; Lewis, Skirton and Jones, 2010; Genetics Alliance, 2010) this may suggest that the findings of my research may be relevant beyond the group of parents participating in the interview study.

One difficulty of the research was the impact of my role as a carer on the progress of the study. As the parent of a child without a diagnosis, I was not fully aware of the risk of recurrence. The difficulties experienced in my pregnancy with my second child, and my role as carer for two disabled children after she was born, meant significant interruption to the research. As data had been collected prior to her birth, and largely analysed, this interruption impacted more on the writing up of the thesis than the interview study. Although being the parent of a disabled child was felt to improve the data collection process in the interview study, it impeded the analysis and writing up process of the study. This delay had drawbacks and benefits. On one hand, it meant my interview study was carried out several years before the thesis was written, and on the other it meant I could incorporate new literature from the fields of the sociology of diagnosis and recent work examining paediatric genetic diagnosis.



A further difficulty of the study was encountered with the intention to share a fairly in-depth account of my own experience of living without a diagnosis. In the end, I included an abridged account of my personal reflections. An important aspect of this decision was protecting the privacy of other family members. Indeed when I read the sections I was intending to include in interludes to my eldest daughter, she objected to the level of detail and I edited and abridged these personal reflections until she was happy with the level of disclosure. This was an important learning experience particularly as, for researchers unlike participants, there can be no degree of anonymity.

### *Contributions to knowledge*

The thesis contributes to and extends knowledge in a number of areas; in empirical work with families; narrative analysis; and to the sociology of diagnosis. Here, I summarise the areas described in the discussion above, suggesting the contribution to knowledge in each field.

#### *1. Empirical work with families*

Key findings resonated with findings in other research work with families with reference to cause, prognosis, access to support and services, and the intensity of the quest for diagnosis. New insights were also made with reference to managing social interaction and making sense of absent diagnosis. Findings in my own work relating to consequences of living without a diagnosis included: that the consequences of absent diagnosis was felt across all temporal domains; that mothers were more likely to express self-blame than fathers; that diagnosis had value in an administrative capacity and was felt to make accessing services easier; that parents utilised strategies for dealing with sustained uncertainty about the future; and that many families reported an unmet need for informal support, which could result in feelings of isolation. New insights were gained in that it was identified that parents may have difficulties making sense of absent diagnosis; non-diagnosis may have low cultural resonance and represent a fracture to parents' assumptive worlds; and parents may continue to seek certainty, despite reducing search activity. The value of diagnostic labels as

interactional tool was identified, as managing definitions of their child in social settings was an unexpectedly common and often intensely felt consequence of absent diagnosis. Isolation parents may already feel, particularly with unmet need for informal support, may be compounded by the difficulties experienced in social interaction.

## 2. *Narrative Analysis*

Contribution to knowledge in the field of narrative analysis was made by utilising and extending Frank's (1995) narrative typologies and by carrying out narrative analysis in conjunction with thematic analysis. Three typologies were identified in parent narratives: *restitution and the new normal*, *times of chaos*, and *unresolved quest*. The latter was identified as the dominant typology. A core metaphor of stasis of movement was also identified, suggesting the difficulty of travel on the parents' journey and/or possibly even representing a contradiction to the dominant typology of unresolved quest.

## 3. *Sociology of diagnosis*

With reference to the sociology of diagnosis, contribution in knowledge was made by careful consideration of the *process* of diagnosis and in considering *genetic* diagnosis. In looking at the process more generally: parents were identified as agentive in the search for diagnosis; parents search activity and thinking about diagnosis commonly decreased over time, although also ebbed and flowed; change (in knowledge or their child) was often the catalyst for search activity; parents gathered, assessed, and re-appropriated e-scaped medicine, with the internet the main tool for their search activity; parents often needed the input of an expert to fully interpret their own findings, although were also seen at times to challenge doctors' own diagnosis work; parents used syncretic understandings to make sense of the diagnosis process, fusing lay with scientific understandings; there was an emotional element to parents' engagement with diagnosis, and a tension existed between the diagnosis search as constructive and hopeful, and also destructive and hopeless.

With reference to the process of genetic diagnosis more specifically: parents practiced a kind of lay dysmorphology, enacting a remarkable level of participation in the clinic; parents

diagnosis work was indicative of the visual culture of dysmorphology; parents sought others in their search for diagnosis in both the context of recognition and belonging; in many parent accounts genetics was played as becoming, an advancing field in the brink of change; the geneticist's own attitude to the field of genetic may have acted as a reference through which the parents drew meaning in the clinical encounter; parents expressed a perception of diagnosis as deferred, anticipating advances in genetic testing; the caring work of the clinic was both medical and pastoral; the clinic was a site of reassurance, a space for hope and connection, an anchor for parents; a process of 'wait and see' was described by parents; a small number of parents considered genetic diagnosis solely through the lens of heredity, whilst a larger number of parents expressed understandings that went beyond explanations of heredity.

I extend the sociological study of diagnosis, suggesting that the absence of diagnosis can tell us much about the meaning of diagnosis; that absent diagnosis opens an analytical window to the possible meanings of diagnosis. Absent diagnosis for uncontested medical conditions is suggested as a new area of diagnostic tension; a topic that is currently under-theorised. Residual categories may not be meaningful for families and the acronym SWAN, like MUS, is a container category that may have limited meaning.

In these areas: the sociology of diagnosis, including studies of the process of paediatric genetic diagnosis; empirical work with families; and in its methodological approach to data collection and analysis, this thesis resonates with existing work and also contributes to knowledge by extending analysis in these fields.

## Conclusion

*On the morning Iris was born, we left for Edinburgh as the sun was still rising, low in the sky. We didn't know whether she would be affected by the inherited condition, although her movement had been normal in pregnancy. Yet so apparently had Maya's. The neonatologist who had cared for Maya, and later Willow, had expressed a 'gut feeling' that everything was going to be fine. I had faith in his tacit knowledge, yet still was anxious. It was a planned C-section, in case she did have the inherited myopathy and didn't survive a normal delivery. The Belle and Sebastian song that Matthew and I had first danced to, before we knew the challenges that lay ahead of us, was playing as she was born. The obstetrician who had cared for me throughout Willow's pregnancy, yet missed her delivery as Willow arrived unexpectedly while she was away, was delivering Iris. The consultant neonatologist who had cared for Willow when she was first born, ventilating her and doing cardiac massage and 'setting her up' in intensive care, was there with his team prepared for the arrival of Iris. It was felt we would know within minutes if Iris had the same condition as her sisters.*

*When she arrived I saw her fleetingly before she was taken over to the team of intensive care doctors and nurses, headed by the consultant neonatologist. I watched, as one by one he sent his team away. Then she was laid in my arms. And another new world was born.*

This thesis has explored the experiences of parents with disabled children living without a diagnosis. Through thematic and narrative analysis of an in-depth qualitative interview study with parents of disabled children, and by considering absent diagnosis in the context of sociological and other relevant theory, this thesis contributes to knowledge about diagnosis and about the experiences of families of disabled children without a diagnosis. In particular, this thesis adds to existing empirical work with families of disabled children who do not have a diagnosis; work looking at the process of genetic diagnosis; and work in the newly emergent sociology of diagnosis. The absence of diagnosis was seen to hold a mirror to diagnosis, exposing the possible *meanings* of diagnosis. It was suggested that the writing of this thesis was, in a sense, also my own narrative reconstruction after biographical disruption. At the beginning of this thesis story I included extracts from poetry and prose written by Ted Hughes and Jacques Lacan. I finish by considering their words and moving beyond them. There is no truth that in the passing does not lie, but one runs after it all the same. I ran, and an absence bounded beside me. I ran, until I could not see an absence.



## Appendix 1

## Participant information table

| Child identifier | Age | Location         | Education setting    | Disabilities as described by parent/s  | Parent interview'd | Method, number and duration of interviews  |
|------------------|-----|------------------|----------------------|--|--------------------|--|
| Tom              | 10  | Central Scotland | Mainstream school    | Learning and behavioural difficulties  | Mother             | One face-to-face interview (95 mins)   |
| Claire           | 6   | Central Scotland | Special needs school | Delays in gross motor, fine motor and cognitive language skills, poor concentration span, reduced sense of danger, chronic heart defect, multiple previous/ongoing surgeries | Mother             | Two face-to-face interviews (90 mins, 70 mins)   |
| Cameron          | 13  | Central Scotland | Special needs school | Difficulties with speech, sleep, understanding and behaviour, and gross and fine motor skills  | Mother and Father  | Mother two face-to-face interviews (2 hours, 90 mins), Father one face-to-face interview (70 mins) |
| Caroline         | 11  | Central          | Special              | Significant difficulties with  | Mother             | Two face-to-face   |

|        |   |                  |                                 |  |                   |  |
|--------|---|------------------|---------------------------------|--|-------------------|--|
|        |   | Scotland         | needs school                    | walking, balance, co-ordination and language. She had no speech and significant learning delay   |                   | interviews (90 mins, 30 mins)  |
| David  | 4 | Central Scotland | Special needs nursery           | Speech, language, understanding and behavioural difficulties, and autistic like characteristics and repetitive behaviours.   | Mother            | Two face-to-face interviews (90 mins, 75 mins)   |
| Emily  | 7 | Central Scotland | Special needs school            | Difficulties with gross motor skills, development delay including very significant difficulties with speech and language and processing information or stimuli, significant difficulties dealing with particular situations, extreme frustration with resulting impact on behaviour, unexplained pain. | Mother and father | Mother two face-to-face interviews (80 mins, 120 mins) and father two face-to-face interviews (80 mins, 45 mins) |
| Olivia | 4 | Central Scotland | Mainstream nursery with support | MRI showed maturation problem with her brain. Delay in gross motor skills, balance problems and significant difficulties with speech, main difficulties speech and communication (with resulting frustration), balance, and difficulties with gross and fine motor skills.                             | Mother and father | Two face-to-face interviews with mother (65 mins, 30 mins) and father (70 mins, 35 mins)                         |
| Kamil  | 4 | Central Scotland | Mainstream nursery with support | Needed a lot of intervention at birth and had several surgeries in his first few years. His difficulties included delay in motor development and speech and  | Mother            | One face-to-face interview (5 hours)   |

|                |          |                     |  |   |   |  |
|----------------|----------|---------------------|--|---|---|--|
|                |          |                     |  | language, feeding difficulties and spinal and other bone structure deformity.   |   |  |
| Andrew and Lee | 19<br>21 | Central<br>Scotland | Special<br>further<br>education<br>centres   | Lee's difficulties - learning disabilities and no speech, Andrew's difficulties - learning disabilities, no speech, walking difficulties and feeding difficulties. They also both had autistic tendencies and a history of hydrocephalus. | Mother  | One face-to-face interview (70 mins)   |
| Ismail         | 9        | Central<br>Scotland | Mainstream school with support               | Sensory issues, hearing impairment, walking and balance difficulties, severe to moderate learning difficulties and global development delay, autistic traits and challenging behaviour.   | Biological sister who was legal guardian/appointed parent | One face-to-face interview (90 mins)   |
| Calum          | 4        | Central<br>Scotland | Private mainstream nursery                   | Growth disorder and learning disability, with particular difficulties with language and communication. He also had sleep issues and development delay in other areas.   | Mother and father   | Two face-to-face interviews with mother (50 mins, 25 mins) and father (80 mins, 25 mins) |
| Abdul          | 4        | Central<br>Scotland | Special needs nursery and mainstream nursery | main difficulties were severe motor development delay, delay in speech and language, feeding issues, and other health issues, abnormal MRI scan   | Mother and father   | Two face-to-face interviews with mother (2 hours, 55 mins) and father (80 mins, 30 mins) |
| Rupert         | 5        | Central             | Mainstream                                   | Attention and behaviour difficulties  | Mother  | One face-to-face   |



|         |    |           |  |  |            |  |
|---------|----|-----------|--|--|------------|--|
|         |    | Scotland  | m school                                   | in the school environment, a vivid imagination, prefer to play alone   | and father | interview with mother (90 mins) and father (55 mins) |
| Peter   | 9  | Australia | Special Needs School                       | Weak muscles and motor dyspraxia, difficulty with lots of physical tasks, a mild intellectual disability, ADHD, behavior problems, health problems, and unable to read or write.   | Mother     | Interview form and telephone interview (60 mins)     |
| Eva     | 7  | England   | Mainstream school with support             | A lifelong disability with complex and moderate to severe needs. She had poor co-ordination and balance, an unusual gait and general hypotonia, fine motor difficulties and some difficulties with speech and social interaction   | Mother     | Interview form and telephone interview (90 mins)     |
| Jessica | 5  | Australia | Not in education and living away from home | Main difficulties were feeding, ataxia, difficulty walking, lack of speech, severe difficulty communicating and very severe behavioural problems, a complete lack of understanding of anything that was said to her, screamed a lot often all night, and bit and hit herself. She had a high threshold for pain and harmed herself often. She had up to 2000 seizures a day. | Mother     | Interview form and telephone interview (2 hours)     |
| Anna    | 11 | USA       |  | Feeding difficulties, hole in her  | Mother     | Interview form                                       |

|       |   |           |                      |  |                   |   |
|-------|---|-----------|----------------------|--|-------------------|---|
|       |   |           |                      | heart, microcephaly and an asymmetric skull. She had difficulties with her eyes and delayed growth in her bones. She had delays in motor development and severe delays in speech and language Mother, interview form   |                   |   |
| Moy   | 2 | USA       |                      | Significant development delay, a series of health and development difficulties on top of feeding and aspiration issues including a worsening scoliosis, hearing and eyesight impairment, and hip dislocation.  | Father and mother | Father completed interview form then mother and father took part in telephone interview (90 mins) |
| Sarah | 4 | Australia | Special needs school | Severe visual impairment and hearing loss, sleep apnea, no speech, asthma, epilepsy, traits of autism spectrum disorder, problems with her hips, feeding difficulties, development delay, respiratory disease, reflux, physical disability, unable to stand or walk. | Mother            | Interview form and telephone interview (65 mins)  |

## Appendix 2 Information Leaflet

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### Thank you for taking the time to read this information

*Should you have any more questions about the research, or if you are interested in taking part in the study, please do not hesitate to contact me by phone, e-mail or letter.*

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*You are invited to take part in this research study. Before deciding whether or not you would like to take part, please take time to read through this information leaflet carefully.*

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#### What is the purpose of the study?

At the moment little is known about the experiences of parents living without a diagnosis for their child's disability. This study will look at the experiences of parents of children with disability, for which there is no diagnosis. It will form part of my PhD thesis.

#### Who will take part in the research?

Both mothers and fathers of children (aged 2 and over) with different kinds of disabilities will take part in the study. Doctors, and others involved in the care of children with disabilities, will also take part in the research.

#### Do I have to take part?

It is entirely up to you if you decide to take part or not. Your child's consultant, or any other professional involved in your child's care, will not be told whether or not you take part in the study.

#### What will happen if I decide to take part?

You will be asked to sign a consent form confirming that you wish to take part. You will be given a copy of this information leaflet and the consent form to keep. Even after signing the consent form, you can withdraw from the interview study at any time and without having to give a reason.

I would like to carry out two interviews with each parent, each lasting about an hour. They will be quite informal and will take place at a time and a place of your choice. Interviews will be tape recorded (if you agree to this) and I will then listen to the tape and transcribe/type up the interview.

In the interview you will be asked about your experience of parenting a child with disability for which there is no diagnosis. You will be given the opportunity to tell your story, and to talk in your own words about yourself and your family and about not having a diagnosis for your child.

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# Syndrome Without a Name?

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## The Experience of Living Without a Diagnosis for Parents of Children with Disabilities

A PhD Study  
**Nicola Coates**  
Community Health Sciences  
University of Edinburgh

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#### Are there any benefits to taking part?

It is hoped that you will enjoy talking about your experiences. I am also the parent of a child with disability for which there is no diagnosis.

Learning more about the experience of living without a diagnosis may help families and health practitioners in the future to understand this situation.

Taking part in the research, however, will not help you to obtain a diagnosis for your child and is not the purpose of the research.

#### Are there any possible disadvantages of taking part?

It might be that discussing some of your experiences may make you feel upset. Should this happen, you will be able to stop the interview at any time and without having to give a reason.

#### Will everything I say be kept confidential?

The names of you and your child, and anyone else you talk about, will be changed in the thesis and other presentations or publications relating to the study. No personal information will be given or made available to anyone else (other than in exceptional circumstances in accordance with child protection guidelines).

In addition to the changing of names, I will make every effort to make sure that you cannot be identified in other ways. Things you talk about that might make it possible for others to identify you, for example the area in which you live or the school your child goes to, will not be included. While it is very unlikely that your participation in the study will come to be known about in this way, it is difficult to guarantee this absolutely.

#### What will happen to the interview tapes and transcripts?

The tape recordings of the interviews will be stored securely and only listened to by myself. The transcribed interviews will only appear in print form with names changed and other personal identifiers removed; only selected parts will appear in the thesis. The supervisors for the PhD: Sarah Cunningham-Burley (University of Edinburgh) and Nick Watson (University of Glasgow), may read parts of the transcripts. You can request a copy of your own transcript if you wish.

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**Consent to Participate in PhD Research Project:****'Syndrome Without a Name? The Experience of Living Without a Diagnosis for Parents of Children with Disabilities'**

I understand that my participation is voluntary and that I do not have to participate if I don't want to.

I understand that I may change my mind about participating in the interview study at any time and without having to give a reason. I understand that I may change my mind about data from interviews conducted with myself being included in the PhD for up to three months following the interviews.

I understand that names and personal details will not appear in the PhD and that, while anonymity cannot be guaranteed, the researcher will make every effort to make sure I cannot be identified as having taken part in the research.

I understand that the PhD, or extracts from it, could appear in conference presentations or reports, academic journals, media articles or other publications.

I understand that my participation in this research will involve one-to-one interviews, which will be audio recorded and later transcribed/typed up by the researcher. I agree to the audio-recording of the interviews and understand that the recordings will be stored securely and only listened to by the researcher.

I understand that personal information will not be passed on to anyone else other than in exceptional circumstances in accordance with child protection guidelines.

**I have read and understood this consent form**

**I have read and understood the information leaflet about the research and any questions I may have about the research have been answered**

**I agree to the audio recording of this interview**

**My signature shows that I wish to take part in the research**

Signature of Participant \_\_\_\_\_

Name of Participant \_\_\_\_\_

Date \_\_\_\_\_

Signature of Researcher \_\_\_\_\_

Date \_\_\_\_\_

Contact details of researcher:

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NOTE: This interview guide outlines question areas for parents of children with disabilities for which there is no diagnosis. Exact terminology and phrases of the questions will be unique to each conversation. The wording is to illustrate areas of conversation rather than exact phrasing of the questions.

**Interview objectives**

To learn about the participant's experience of parenting a child with disabilities for which there is no diagnosis.

To explore the parents' perception of and attitude towards diagnosis and the situation of non-diagnosis.

**Section 1: Prompt a narrative history**

What I'd like first of all is for you to tell me your story. I'd like you to tell me about yourself and your child. You can start wherever you want: for example when your child was born or when they first started to have difficulties, it's up to you, and it can be as long or as short as you like. I'd just like to hear your story however you want to tell me it, and to learn a bit more about you and your child.

*NOTE: It is expected that in the narrative history section the parent will raise issues relating to diagnosis/non-diagnosis. Issues raised will help to shape the rest of the interview.*

**Section 2: Prompts to elicit more information about the child and the situation regarding diagnosis**

So how would you describe your child's difficulties (*recap what they've already mentioned*)?

Thinking more specifically about diagnosis now, can you tell me about what's happened in terms of diagnosis for your child since it was first obvious that they were having difficulties?

Do you know what kind of testing or assessments have been carried out so far?

And what about now, what's the situation at the moment in terms of diagnosis for them?

### **Section 3: Prompts for exploring possible effects of non-diagnosis and attitudes to diagnosis with the parent**

Has not having a diagnosis for your child's difficulties had an effect on your life do you think?

*(if yes)* In what way do you think not having a diagnosis may have had an effect on your life?  
*(further prompts)* What about practically? What about emotionally?

Do you think that not having a diagnosis might have had an effect on your child?

*(if yes)* in what way/s?

Do you think not having a diagnosis has had an effect on your family life?

*(if yes)* in what way/s?

I'd like to talk now more about how you think not having a diagnosis may have had an effect on you and your child's lives. I'd like to think about different areas in your lives and whether or not you think not having a diagnosis has had an effect on these areas *(acknowledge that they have already mentioned several of these if appropriate)*.

I'd like to ask you if you think not having a diagnosis has had an effect on the following areas:

*(prompt to discuss not having a diagnosis with reference to the following areas)* Cause, prognosis, access to informal support, access to formal support and services, ability to cope with and adjust to disability, how to describe their child's disability to others, future family life and family planning.

*(In each case define the terms if necessary and prompt for further information as appropriate)*.

I'd like to ask you now a few more questions about how you think not having a diagnosis may have had an effect on you.

Are there any other areas of your life, other than those areas we've already talked about, that you think not having a diagnosis might have had an effect on, either practically or emotionally?

How do you feel about not having a diagnosis?

I'd like to ask you now about how you think about diagnosis and about your child not having a diagnosis

How much would you say you think about not having a diagnosis?

Do you think you used to think about not having a diagnosis more, or less, or just about the same?

Why do you think that diagnosis has not been made for your child?

How likely do you think it is that your child will get a diagnosis?

Who do you see as responsible for diagnosing your child?

What, for you, is a diagnosis? *(can prompt examples if necessary)*

What, for you, is not a diagnosis?

How important do you think it is for a child to be given a diagnosis?

What benefits do you think having a diagnosis might have for you and your child?

Do you think there are any negative sides to having a diagnosis?

Do you know any other parents of children with no diagnosis for their child's difficulties/disability?

Had you ever heard about children with disabilities with no diagnosis before you experienced not having a diagnosis for your own child's difficulties?

#### **Section 4: Prompts for discussing the diagnosis process**

I'd like to think more specifically now about the process of diagnosis, and how you have experienced this process.

Do you feel that you yourself have looked for a diagnosis?

*(if yes)* In what ways do you think you've done this?

*(if yes)* Do you think that the way you have looked for a diagnosis has changed over time?

What might start or stop you looking for a diagnosis?

Can you think of anything that has happened that has made you start or stop searching for a diagnosis?

Thinking now about the way that you were made aware that there might not be a diagnosis for your child's difficulties.

In what way was this explained to you, if at all?

*(can prompt if necessary: by whom, when, context, timing, info given)*

Do you think it was explained well to you? *(prompt reason)*

Is there anything that could have been done, do you think, to have made it easier for you to understand the situation?

**Section 5: Prompts for discussing parents' attitude to medical practitioners and perception of medical science.**

I'd like to talk a bit now about your views on doctors and other professionals involved in your child's life.

How do you feel about doctors?

*(prompt with reference to different fields/levels of practice, eg: GP, pediatrician, consultant)*

*(prompt reason)*

Do you think not having a diagnosis for your child's difficulties has had an effect on the way you feel about doctors or professionals involved in your child's life?

Final prompts for ending the interview

Is there anything else you'd like to say about diagnosis or the situation of non-diagnosis?

Do you have any questions you'd like to ask?

How have you found this interview?

Would you like me to send you a copy of the transcript of this interview?

*Thank them for taking part.*



- What's been happening for you since last interview?
- One of the things you told me about in our last interview was the way that the doctor described Emily when she was first born as having a number of features they thought were unusual [features removed for confidentiality]. Can you say just a little more about that early experience? How did that feel?
- In the first year of Emily's life, you talked about the stressful screaming at night. When was it first clear that there may have been something wrong and that this situation was different to the experience you had with your other daughter having very bad colic?
- How did it feel when you found out it was your *father* who was the [name of syndrome] carrier rather than your *mother*? How did you feel about your sister having been told previously by [name of Dr] that it wasn't possible to have a 'normal transmitting male' ie your father?
- You said it was realised eventually that the doctor was wrong about your father not being able to be the carrier and that this showed how 'things [medical knowledge] progressed over the years'. Do you think you compare that situation to the situation with Emily not having a diagnosis? – in that you said that although they 'haven't a clue what's the problem at the moment ten years down the line they might'.
- You talked about the behaviour that you called 'name of behaviour' that you associated with [name of symptom] but the health professionals had a different opinion about. Has there been any more discussion about what this with the health team? Do you feel that if she had a diagnosis this might be more likely to be explained? How does it feel to disagree with the doctors and consultants about this?
- How did it feel when [name of consultant] 'made more sense' of everything and was able to talk about why Emily, for example, found supermarkets difficult etc?
- You described the clinics you've been to before with Emily when diagnosis has been discussed as a 'waste of time' – can you say a little bit more about that?
- You said the appointments with the geneticist have 'finished' – just to check, is Emily seen at all by the geneticist now?

- You described how, according to the tests, Emily is 'the most normal child you could get' as nothing has shown up with the tests. How does this feel?
- One of the main difficulties you described with not having a diagnosis was what to put on benefits forms. I just wanted to confirm that you felt that not having a diagnosis made that process much more difficult?
- Another of the main effects you described was problems with trying to explain to people what was wrong. You said you felt it was difficult to describe her difficulties to others, and that saying she had special needs didn't 'allow people to understand' and you felt it was too much of 'a wide spectrum'. You also said you felt it would be easier if she had a diagnosis like Down syndrome and that it was difficult at the moment without going into lots of details to communicate to others what her difficulties were. Could you say a little more about that?
- You felt that it would be difficult to get support from other parents/join support groups and even though that wasn't something that you yourself might be interested in, you felt [name of Emily's mother] would. Can you say a bit more about this idea that you get groups for Down syndrome, groups for autism but that when you don't have a diagnosis you [in his words] 'don't fit into any sort of wee pocket anywhere'?  
Do you think that parents of children with Down syndrome *do* fit into 'a wee pocket somewhere' – and how?
- You said, with reference to the cause of her difficulties, that it would be 'nice to know' but it wasn't something that 'bothered you as much' now and you wouldn't be thinking along the lines of 'apportioning blame' – do you want to say anything more about that?
- Could you say a little bit more about how you think not having a diagnosis may have contributed to the way you see the future as uncertain?
- Do you think that not having a diagnosis for Emily makes it more likely that people may not believe you about her difficulties, for example if they meet her when she's in [what he called] a 'good mood'?
- How much do you think the [name of syndrome] experience with [name of relative], and the idea that '30/40 years ago they wouldn't have known what was wrong with him but now they do', has an effect on the way you think about Emily's diagnosis situation?
- Can you say a little more about your opinion that 'genetics is a great thing but it can be a dangerous thing as well'?

- You said you felt 'let down by the system' because they're not 'able to tell you what the problem is when there quite clearly is something wrong' Can you say a bit more about feeling let down?
- You described how you felt that 'things are always progressing as the years go on' and how, in your words, you 'don't think for one minute that we'll never have a diagnosis' and that 'I think we're eventually going to have a diagnosis at some point in life'. Can you say just a little more about this?
- You said you felt there's no point in dwelling on things in life, what do you do instead with things you might find difficult?
- You said you 'think that it's a genetic thing', that you're 'quite positive' it's something in the genes' but that the scientists/medical people 'don't *know* what the genetic problem is yet'. Why do you think you feel so strongly it's a genetic thing?
- With reference to your feeling that you're 'quite sure it's come from either yourself [was myself] or from[name of Emily's mother]'. Can you say a bit more about that? Do you speculate about which side of the family?
- How much do you think 'how much you push for it' will influence whether or not you get a diagnosis?
- Can you say a little bit more about how you see the phrase developmental delay as a 'cop out'?
- How related do you think having a diagnosis is to getting the right treatment or services for Emily?
- You said when you've got a diagnosis 'you know what you're dealing with' – could you say a little bit more about what you mean by this?
- You mentioned you 'wouldn't like to get a label stuck on her just for the sake of putting a label on her' – can you say a little more about this?
- You talked about your [name of relative] having severe learning disabilities and how nobody ever knew 'what the problem was' and it was 'put down to him being dropped as a baby'. You mentioned that you'd thought about the possibility that that might be exactly what Emily's got – could you say a little bit more about that?
- Do you think that your [name of relative] telling the staff he had a [name of symptoms] and them not believing him had any impact on how you feel about Emily with her problems with her[name of symptom]?

- How do you feel about the consultants not being able to come up with any answers about her recurrent [symptom removed for confidentiality]?
- You talked about [name of Emily's mother] coming up with a name for Emily's auditory and visual processing difficulties and how you felt that it 'wasn't an official diagnosis' but it was 'pretty close to the way Emily actually is' – can you say a little bit more about that?
- You talked about how 'new things are being found all the time' in medical science/genetics. Can you say a bit more [about what kind of new things]?
- You said some of the doctors you've met over the years – 'think they're god' and that they're 'up on some kind of high pedestal somewhere and whatever they say goes' – can you say a bit more about that?
- You expressed how you felt that 'doctors don't know everything' - Can you say a little bit more about that idea?
- You said in the first interview that 'there might be a diagnosis some day but at the end of the day it's not going to be the end of the world if we don't get one'. Would you say this summed up how you feel about not having a diagnosis? Is there any other way you would like to sum up the situation of not having a diagnosis for Emily?
- Do you have anything else you'd like to say about not having a diagnosis?
- How have you found the questions?.....etc

- Acceptance of and coming to terms with disability
- Access to formal support and services
- Access to informal support
- Alternative knowledge systems
- Ambivalent feelings towards dx (diagnosis)
- Attitude to residual~umbrella cat's
- Cause
- Child's appearance~inc dysmorphology
- Current diagnosis situation
- Deferral
- Disclosure that there might not be a diagnosis
- Doctors given up on dx
- Down syndrome and autism non-spontaneous
- Down syndrome and autism spontaneous
- Effect of non-dx on insurance claims
- Effect on child of non-dx
- Effect on parenting
- Effect on sibling
- Effects of non-dx ~spontaneous~
- Epiphany moment in narrative
- Extended family difficulties
- Future family planning
- General comments about diagnosis
- Gold Quotes!
- Has non-dx effected how they see drs-medical science
- Hope
- How important is or benefits of dx
- How likely they think dx is
- How much they think about non-dx
- How they feel about doctors
- How they feel about non-dx
- How to describe child's disability
- Impact of 2nd baby
- Knowledge of genetic~testing process
- Knowledge of non-dx or other parents
- Metaphor
- Missed health problems due to non-dx
- Negative sides to having a diagnosis
- Negotiating dx with med prof
- Non dx conceptually hard to deal with
- Not being believed due to non-dx
- Parent disagreeing with medical opinion
- Passing of time
- People's expectations of child due to non-dx
- Personality affecting experience of non-dx
- Personality~pathology boundary

- Positives to not knowing
- Prognosis
- Prolonged testing
- Purpose and place of the clinic
- Preference to media
- S~W~A~N~
- Suggest why they think dx not made
- Their process of looking for a dx
- Travel insurance
- Treatment-cure
- Trust in medical encounter~science
- Unable to research as effect of non-dx
- Uncertainty~living with sustained
- Uniqueness~anomaly
- What is a dx
- What is not a dx
- Who they see as responsible for dx
- Why they think dx has not been made

**Syndrome Without a Name – support organisation**

This Appendix includes information about the support organisation SWAN, then considers data from the interview study with relation to contact with and awareness of SWAN, before discussing the interview I carried out with the founder of SWAN who, at the time of the interview, ran the support organisation.

Syndrome Without a Name (SWAN) was set up in 1999 by the grandmother of an undiagnosed child. Some of the parents in my study had knowledge of SWAN, and/or experience of accessing the organisation's website or of making contact with families through the organisation. In 2007, I interviewed the founder of SWAN at her home in Staffordshire; where the organization was based at the time of the interview study. Between 1999 and 2009 SWAN provided support and information to numerous families, with membership peaking at 1300 families. Links were formed with a number of health and social care professionals in the UK and with similar support groups abroad.

When the founder was no longer able to continue the group, it became inoperative for two years (2009-2011), during which time the charity Genetic Alliance UK became involved in taking on responsibility for the future of the group. When SWAN was re-established as a support group again in 2011, its purpose and the way it was co-ordinated had changed. No longer run by the family member of an affected child, SWAN UK was run by Genetic Alliance UK, with a mission statement of 'offering support and information to families of children with undiagnosed genetic conditions'. Perhaps indicative of the increasing geneticisation of explanation for disease and disorder, the support offered to families was now assumed to relate to families of children with conditions with genetic aetiology.

In 2010 Genetic Alliance UK secured funding to carry out preliminary research with families to identify their priorities for the future work of the charity SWAN. This work was used to evidence the need for a specific supportive service as the difficulties identified by families were seen to be unique to the situation of living without a diagnosis. In 2011 Genetic

Alliance UK secured funding from the Big Lottery Fund to employ a full time coordinator for 5 years to re-establish the SWAN support group. Based on the needs identified during the research consultation, 3 aims were specified: to develop a community of families with undiagnosed genetic conditions for mutual support and information sharing, to develop a network of health and social care professionals with expertise in undiagnosed conditions, and to increase awareness and understanding of undiagnosed genetic conditions. The support work initiated by Staffordshire Grandmother, therefore, was continued in a more formal way, coordinated by Genetic Alliance UK and with an advisory committee overseeing the work of the charity. A number of parents of undiagnosed children are members of this advisory committee. The focus of the group is also more specific than it was at the time of the interview study, with support offered to families of children with unidentified genetic conditions.

At the time of the interview study for my research, membership was open to all families without a diagnosis for their disabled child. Only three of the parents in my research knew about the support organisation SWAN in the UK. Eva's mother and Abdul's mother and father were the only parents to have used the support services of SWAN UK. While Abdul's father had only 'had a look' at the support organisation's website and occasionally read over the SWAN newsletters online, Abdul's mother had made quite extensive use of the organisation's website and had made contact with other families via SWAN. She described the benefit of making contact with others who were also parents of children who didn't have a diagnosis.

'That's kind of nice because you feel like...you have this connection with somebody...as soon as you make contact you do have a connection because you have this big thing you have in common and it means that you know there's so much you can talk about that they just identify with straight away'

She described making contact with others who posted their details on the website, as well as putting her own story in one of the online newsletters. She recalled how she did this at a time when she was feeling low; 'desperate for some contact'. She found this beneficial as she received a steady stream of responses to her story, although none from families in her geographical area. Contact was made by families who saw some similarities between Abdul



and their own child. In posting her own story, and reading through the stories posted by other parents, Abdul's mother was looking for families with children with the same or similar clinical characteristics to her son. She also felt she received beneficial advice from some of the people she made contact with, including comments on whether to go ahead and have another child. She recalled hearing the phrase 'syndrome without a name' from her child's geneticist and liking the way the phrase sounded.

'He was the one who sort of explained to us about this idea of syndrome without a name, so that's what we do when we find out about something you go to google, see what you can find...some of these terms are so horrible that SWAN sounds you know really nice.'

She felt, however, it would have been helpful to know about SWAN earlier and useful if parents could be given a leaflet about living without a diagnosis early in their journey if diagnosis seemed unlikely.

Eva's mother also regularly visited the SWAN website, often reading the posts of the other parents. She discovered the site by accident when looking online for information about diagnosis; 'one of my searches took me there once'. She was keen to find another child who had similar difficulties to Eva's but didn't manage to make contact with another family with a child like Eva.

In addition to these parents, four of the parents who took part in the interview study knew of the SWAN support group in their own country (one in the USA and three in Australia). Two of these parents (Anna's mother and Sarah's mother) had set up the support organisation in their country after being inspired by the SWAN UK group. The USA and Australian SWAN groups are run by these parents and have similar mission statements to that of the original group in the UK, aiming their support services at families with an undiagnosed child without specifying a genetic link.

Sarah's mother had set up the SWAN support organisation in Australia in 2003 because there was no such support group in Australia. In her role as founder of the organisation she maintains contact with families, organises family meet ups including a family retreat, raises funds and seeks to raise awareness of disabled children without a diagnosis. Awareness is

raised at a local level, for example, in schools, as well as at a government level. She said in her interview that she aimed to provide support, motivation and empower families offering them opportunities to have a voice. Sarah's mother used the phrase 'SWAN child' to describe her daughter and encouraged doctors to do the same. She said;

'They [doctors] ask me what it means I use the phrase to make them think that things aren't always as they seem like the story of the baby swan who everyone thought was ugly and grew up to be quite beautiful'

She told me that her daughter's file at hospital had the phrase 'SWAN child' written on the front of it, at her request.

Anna's mother set up the SWAN support organisation in the USA. This strand of the organisation aims to provide support for families offering information, understanding and empathy and empowering families to advocate for services for their child. They also advocate for increased awareness by the medical community, education system, social services, and the general public about undiagnosed children; improved care; and improvements in the diagnosis process. Part of this latter aim involves striving to set up a database in order to make diagnostic connections as well as featuring children without diagnosis on their webpages so that others who may have any information regarding diagnosis or who may have a child with similar characteristics can be in contact with the families. Anna's mother said she set up SWAN in the USA 'to help support those who feel alone'. Like Sarah's mother, she was inspired by the work of SWAN in the UK.

Jessica's mother and Peter's mother (both from Australia) were also familiar with SWAN. Jessica's mother knew of one other family of a child without a diagnosis through contact via SWAN, although their children were very different ('her son is chalk and cheese with my daughter'). In terms of the face-to-face support from the organisation, Jessica's mother had found it difficult to access meetings due to geographical distance. Peter's mother had only just found out about the support organisation SWAN. Since discovering the organisation she had read stories of other families in the hope of finding a child who resembled her own; 'I have heard about SWAN and read the stories of other parents in the hopes that I might see

someone similar'. She felt it would have been beneficial if the doctors involved in her son's care had known about SWAN and had put her in touch with the organisation earlier.

Very few parents mentioned the support organisation Syndrome Without a Name (SWAN) when talking about access to support, although most described unmet need for support. Making contact with other parents of children with similar difficulties was clearly desirable for many parents. Of those who had made contact with families via SWAN, one parent described making valuable contact with other families, although no parent felt they had identified other families with children with very similar difficulties.

#### *Interview with Liz; the founder of SWAN in the UK*

An interview was carried out with the original founder of the SWAN support group in the UK. Here, I include data from our interview for the purposes of greater understanding of the support provided by SWAN at the time of the interview study, and also as a form of triangulation of the themes that emerged from the interview study with parents. Liz had unique contact at that time with parents living without a diagnosis and a good understanding of their perceptions of absent diagnosis. She met with parents, had email and written communication with them, and personally manned the SWAN helpline that parents called for support and advice. She also had direct experience of a family member being severely affected by a disability that had no conclusive diagnosis. As she was not the parent/guardian for a child without a diagnosis, I did not include the data from her interview in the data analysis elsewhere in the thesis, but I felt it was important to acknowledge the issues raised in our interview. Our interview consisted of several parts and I constructed the semi-structured interview guide with the aim of finding out more about the support organization SWAN; hearing her own story about her experience with her granddaughter; and learning more about the issues parents came to SWAN for support with.

Our interview, which was carried out in her own home in Staffordshire from where she ran SWAN, began with Liz describing why she set up the support organization. Liz set up SWAN following the birth of her grand-daughter. As the grandmother to a child without a

diagnosis she embarked upon trying to make contact with other families in the same situation or with children with similar difficulties. She was put in touch with several families by the support organisation Contact a Family. Soon she found she had a small database of families she was in contact with and decided to set up a 'small local group' offering these families support. The plan was to meet every month and have a coffee. The group grew, however, and soon she began making contact with families further afield. Contact a Family helped the group grow and continued to put Liz in contact with families of children who didn't have a diagnosis. Information about SWAN was disseminated through the press and radio. Soon, Liz was making contact with families and organisations nationally and internationally and the membership of the group was growing. Health and social work professionals began to refer families to the group for support.

At the time of the interview, the organisation had over 1300 members on their mailing list in 7 countries; with around 1200 of these members living in the UK. Liz manned the phone line (her own landline) for the organisation and worked along with several volunteers to keep in contact with families via a monthly newsletter. At the time of the interview, there was also a website for the organisation, with a forum for parents to make contact with each other and discuss topics of interest. Liz had previously put families with children with similar difficulties in contact with each other. She also provided families with information or referred families to other organisations that may be able to help them. The organisation worked to raise awareness of children without a diagnosis, at local and government level, and a main part of the mission statement was to generate a database of undiagnosed children so that they may be matched with diagnoses.

During our interview Liz was invited to tell me her own story of being the grandmother to a child without a diagnosis as well as to share the knowledge and experience she had gained from running the support organisation SWAN. Liz identified issues faced by others parents, as well as the issues faced by her own family, in the absence of diagnosis. These issues were consistent with findings from my own research with parents and with other studies with families with disabled children who did not having a diagnosis. The interview with Liz, therefore, offered some triangulation of results of the data generated by the interview study with parents in my research.

Liz felt that not having a diagnosis had an impact for parents on access to formal and informal support. She reported that many parents had spoken to her about difficulties making contact with other families. Parents had also identified that other families with a diagnosis had more information about cause and prognosis. She identified feelings of isolation for parents of children without a diagnosis who may not 'fit into a particular slot'.

'A lot of parents have said to me we go to a support group but everybody there knows what's wrong with their child and they can talk about what caused it and the prognosis We can't We feel left out and a lot of parents have said that to me you don't fit in to a particular slot and again there's always this perception of what medical science can and can't do (...) and they do say that that there's this awful feeling of isolation'

Here, Liz identifies that access to an appropriate support group is difficult for families without a diagnosis and the isolation that lack of support can bring. She also hints that parents may have expectations of medical science that are not met and highlights how cause and prognosis may not be known in the absence of a diagnosis. She reports a perception among parents without diagnosis that other parents of children with a named diagnosis have more information about cause and prognosis. Access to formal support and services was also raised as an issue for parents, including access to benefits, gaining the right educational support and accessing equipment.

'Benefits again is a major area of concern because if you don't have a diagnosis then you've got to prove that you have a need Education a major area of concern (...) because there's no guidelines the children are so different and they don't fit into any category getting equipment like a wheelchair or a buggy again the onus of proof is with you you know you've got to be prepared to fight for everything and to see children who have fewer disabilities get them relatively easily it is a major area of concern'

She also spoke of difficulties for parents with assessing the risk of recurrence without a diagnosis, another theme identified in my own study and in other work with families with undiagnosed children.

'[unknown risk or recurrence] is a problem and I do know that some families they've gone on to have another child which I think is terribly brave and the child had been healthy some sadly the child hasn't been but without that definition (...) there's no test for it you've got to take your courage in both hands and say yes we're going to risk this'

Regarding unknown aetiology, Liz also spoke of encountering families where the stress of not knowing what had caused their child's difficulties had impacted on relationships. She felt this was 'one of the major problems the families face' and said; 'it's got to be difficult when you don't know why you know who caused it is it the mother? is it the father? is it both? is it none? is it just a quirk of nature?' She reported that some of the mothers who had been in touch with her had been blaming themselves for the child's difficulties.

Liz talked about use of the phrase 'swan child'. She had been in touch with the Royal College of Pediatrics about gaining more formal recognition of this phrase, and showed me their reply letter in which they explained that it wasn't possible for this phrase to be accepted as a diagnosis. Liz explained to me that she wanted the phrase to become an 'accepted term'; 'it would mean there is a genuine case here it just simply doesn't have a name'. Liz felt the phrase was 'handy' and it was a 'gentler way of explaining the problem rather than saying you don't know'. She reported that 'parents have said it does give them a feeling of belonging to a group and its identity'. She identified that for parents without a diagnosis, knowing how to describe their child's disability to others and gaining a sense of belonging to a particular support group was difficult.

*The Story Lisa Told Me While Kelly Ironed Her Hair*

I had my daughter to a different person  
Then I met Claire's dad and I got pregnant but I miscarried  
And we just put it down to miscarriage  
Then I fell pregnant with Claire  
  
When I went for a routine scan  
They said her neck was thicker than normal  
Which indicated some kind of syndrome  
And then they put it down that it might be Down's syndrome  
  
They tried to bully us into a termination because they kept phoning  
And Philip said look!  
Will you just leave us alone  
We're not having a termination, we're having a child  
It was like well there's enough children with special needs in the world  
We dinnae want another  
That's how it felt to us  
Over the next couple of weeks they kept phoning  
We just carried on the pregnancy  
  
She was two weeks late  
And when she was born they were 99.1% that she was Down's  
Because she had all the features  
She had the neck, the bridge of the nose, her ears wernae matched  
The one thing she didn't have was a crease  
And they have a crease right across their palm  
But she didn't have a crease

They done chromosomes  
Then they asked for the chromosomes test again  
And they found out that she wasnae Down's

She must have been about a month old  
When they told us she wasnae Down's  
We had come home thinking  
That she had Down's and a heart problem  
I knew a wee bit of Down's  
Cause my friend's wee lassie was Down's  
And I've seen her grow up since she was two

Well they done mine, Philip's and Kelly's chromosomes  
And they couldn't see  
So they said we don't know what she's got  
Because they've done loads and loads of tests  
The geneticist done god knows how many tests  
All the tests that were available  
And they still came back that they didn't know what she was  
I was relieved really relieved  
But then we came to, well what has she got?  
And we're still at that [laughs]  
Seven years later we're still at that stage

She didnae walk until she was four  
She used to use a wee zimmer  
And the doctor had noticed her hips wernae matched  
And that's what made him think of GYRT Syndrome\*  
But we looked up on it  
And we couldn't understand  
Because all them we'd seen with this GYRT Syndrome



Didn't have the heart problem  
And we spoke to him about that  
And even he said he wasnae happy that she was in the category of this GYRT Syndrome  
But they were classing it just now  
Basically she's still at the stage when she's got an unknown syndrome

[long pause]

She had to have her first heart operation  
They gave her a 50-50 chance  
We had to sign the consent form wondering  
If we dinnae sign it she could still have time with us  
And if we do sign it she could die anyway  
Basically she was going to die anyway  
So we gave her the chance of the operation  
Luckily it was a success

We thought it was maybe just a one off thing with her  
But my sister had fell pregnant with a little boy  
That's him up there  
And he had a heart condition as well  
And the doctor thought it's just co-incidence  
My nephew em [pause] died a couple of year ago  
He was only just turned two  
Yeah he died because his heart  
It wasn't only his heart that was affected  
But it was like five different things  
There was bits back to front

Before he had died I'd fell pregnant again  
With a wee laddie  
But em at twenty-two I kept bleeding

So we went up and at twenty-two and a half weeks  
They says he didn't have any kidneys  
And he said babies born without kidneys willnae survive  
So I had to have a medical termination  
And that's when me and my sister  
My sister's looking into my mother's side  
Well my mother's father and my mother's mother's side  
Just to see if there's any  
Because it's more than just a coincidence  
There's something doon the line  
There's something that's just not adding up

(...)

It's been a worrying time  
But she gets through it all the time  
She's had her second operation  
She's nothing but a worry spot that one  
A lot a lot of hard work

(...)

They even took an x-ray  
Because the doctor says now and then  
A load of doctors from different countries all meet in England  
So he put her case forward  
But nobody else had seen it

We don't know  
She's still undiagnosed  
I'm not really bothered anymore  
I would like to know

It's hard to tell people  
And people say what's wrong with her?  
How can you tell them as you'd go into all the details  
You can get frustrated with people  
When they ask what's wrong  
Oh we don't  
How do you not know?

He's told us there's nothing available  
There's nothing more available that they can do

It's just not knowing  
If we could just have a name that we know definitely  
That we could go  
It'd make things a lot easier  
I suppose like for peace of mind  
You'd just like to know what it was your child has

My mum gets confused as well  
She'd like to know as well

If we had a diagnosis then maybe they would know what to do

I would like to know why  
Maybe if we knew how  
With each side of the family [laughs]  
Whose fault it was [laughs]

I've always blamed myself for the way she is  
And her dad blamed himself  
Because I was the one that carried her

And he feels it was his fault because he was the father  
Cause I mean I never  
I did smoke when I was pregnant yes  
But I never drunk any alcohol  
I never took any tablets or anything like that  
But you just feel because you're the one who's carrying them  
You should be the one that's protecting them  
And when they've got something wrong...

[pause]

We don't know much about my father's side  
So we don't know if there's something on his side  
Cause his son's got an eye defect  
But I don't know any more about that  
We think it's definitely my side of the family

We were relieved it wasnae Down's  
We were relieved  
But then it came to the unknown syndrome so we just  
Every time we thought we had something  
We ended up just back to square one again  
It feels like a roller coaster that's never going to stop

They cannae give us a lifespan on her  
They could say oh she's got one year left she's got ten years left  
We just don't know  
And that does make it hard  
At least when you've got a diagnosis you could look it up and think  
Oh well that person's there lived to that age  
And you could have a bit of an idea

That could be the hard bit

Not knowing

It's hard to try and describe what she's got

It is, very difficult

Because if Claire was Down's you'd say

Oh she's Down's

And a lot of people, the majority, know what that's like

They can accept that

But see you say they think she's got an unknown syndrome

They just look at you

It is frustrating

At first it was upsetting

Wondering

You wanted to know what's with your child

But cause it's been like nearly seven years

It doesn't bother me anymore

I'd still like to know what it is

But it's not like a major thing now

I still think about it all the time

More than likely every day because I see Claire aye

(...)

I don't know why a diagnosis hasn't been made

Cause they've done all the chromosomes and everything's normal

She's no got any extra or any more

They've done everything they can possibly do

They've counted her blood cells and everything

They've done every test that's available

The nurse at the hospital used to call her 'Claire no syndrome'

It could be something me Philip's got  
And it's one in every thousand that could meet up  
It could be genetic  
They just don't know  
Cause they done all our chromosomes  
I think they were going to look into the genes or something  
But there was no more equipment available to do any more testing

They say there's not another child  
That they know of  
In the world with what she's got  
It gives me goosebumps when I think of that  
Cause surely there must be other children the exact same as her  
But they can't find any  
I wouldn't want to be the only one  
That my child's got something different  
From every other person in the whole wide  
in the whole of the population

We'd like a diagnosis  
That would suggest everything that was wrong with her  
Then it would start to make sense  
Just knowing, at last

We got some information off the internet  
But a lot of it was like doctor-words  
So me and my daughter went to the library  
And got a book out there  
And read what the actual words meant  
It was just a book on syndromes

Where we found out what the actual names meant  
If they told us there was something else then we'd look up  
We've looked up all what her problems is  
We've tried everything and nothing comes up that's got aw her problems  
Some of her problems yeah but not all

It was Paul's brother's sister  
No 's Paul's wife's sister so Claire's auntie  
Had put a website up for her on the computer  
Just asking if anybody's got any similarities  
And we went and me and Paul done some study ourselves  
Went through the computer  
Me and Kelly went to the library with all the names and all that  
And it just didnae add up  
That's when we spoke to the doctor  
That's when he said he agreed with us  
He said he wasnae happy with it either  
So he must have obviously been discussing it as well

Some doctors are all right  
But the doctors speak in these big fancy words  
Ah mind them saying that she wasnae well, she wasnae well  
And her body had stopped growing  
And they were worried in case her brain was going to outgrow her head  
And that was in the consultant room  
And I mind coming out thinking her brain's just going to go poosh!  
And I spoke to a nurse  
And the nurse explained it which made it a lot easier  
The doctors use the big fancy words  
So far better asking the nurse  
  
I ken they're no miracle workers

They do try  
I do admire doctors I really do

I mind at first we were really upset  
And paranoid really  
Because we just really wanted to know what was wrong with our child  
I'm not as bothered anymore  
But I'd like to know, just personally aye but

I've started to think about it  
More and more aye  
It's just I read all these magazines  
People that's got children and they find a cure  
They find a name and thingmy  
And I just think God I wish I could find a name for mine  
I didnae realise how much I actually think about it  
I keep thinking we're going forward  
And we keep going back  
It's still a rollercoaster  
Some pit stops along the way and that's it  
If they gave us a word  
We'd be able to look it up  
Each different bit  
And we'd be able to put it together and see what her future holds  
See what she's going to be capable of

I think it's just  
Cause if I knew it was definitely my side  
Then Paul's got a new girlfriend now  
He could go for a baby with her  
He's scared to do it at the moment in case something happens

I don't think that the answer is there



I don't think they'll ever find out  
Cause I just think it's one of these things  
They know the name for each thing that's wrong with her  
They've just not got a name for the whole thing

You feel frustrated at times  
Sometimes it doesnae bother me  
And sometimes it feels frustrating  
I'm still looking

[long silence]

Probably  
If we had some sort of name for it  
Or something even sort of some recognisable name  
Or something  
Then I could find out about my father's background  
Maybe there's something there

[long silence]

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